



Barth Syndrome
Foundation

www.barthsyndrome.org

Barth Syndrome Journal

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



Photo courtesy of Amanda Clark - 2010

Great News about Barth Syndrome Individuals' Longevity

By Kate McCurdy, Board Member, Barth Syndrome Foundation

For several years, the Barth Syndrome Foundation (BSF) has made the claim that individuals with Barth syndrome (BTHS) are living far longer than ever before. Now we are delighted to have the data to prove it. **All of our collective efforts ARE making a difference!**

BSF was incorporated back in 2000, and by the end of that year, we had 41 living boys in our group who had been diagnosed with BTHS (see *Graph #1 on page 4*). As the literature indicated then, most BTHS individuals died very young, and, indeed, you can see that fewer than 10% of those we knew living then were over 15 years old. That year, however, the oldest BTHS individual in the world of whom we were aware was in his forties, and that gave us great hope.

Compare these figures to the similar ones today (see *Graph #2 on page 4*). Our organization has grown to include 148 individuals from around the world living with a genetically confirmed diagnosis of BTHS. Now, more than 35% are older than 15 years old. Furthermore, the oldest man living with BTHS whom we know of now is in his sixties!

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A Continuum of Collaboration Barth Syndrome 2012 Scientific and Medical Sessions

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



Photo courtesy of Don CeSar Hotel
Venue of BSF's 2012 Conference

Most attendees of the Barth Syndrome Foundation (BSF) International Scientific, Medical & Family Conferences are not aware of how the Scientific and Medical Sessions (SciMed) are put together, so I would like to take this opportunity to explain about this important part of our Conference. From the very beginning, these biennial BSF-sponsored conferences have combined the families with the physicians and scientists who all want to find a better future for Barth syndrome (BTHS) individuals. As the Conferences have grown in size, the number of physicians and researchers wanting to attend these Conferences has grown as well. There is little doubt that the success of the BSF Research Grant Program encouraged a lot of this growth, particularly among the scientists for whom the Conference has become a prime opportunity to communicate the results of their research. The 2012 SciMed sessions of the Conference will be no different

and will continue with the tradition of providing the most up-to-date results about scientific and clinically-relevant research done on Barth syndrome.

(Cont'd on page 5)



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Letter from the Chairman

By Stephen B. McCurdy, Chairman, Barth Syndrome Foundation

The Barth Syndrome Foundation (BSF) Board of Directors is thrilled to announce the appointment of **Lindsay Groff** as Executive Director. Lindsay brings a variety of experience to BSF, including nonprofit management, conference planning, marketing, and the perspective of a parent advocate associated with the Children's Hospital of Philadelphia (CHOP). We spent months searching for the right person and are delighted to have found Lindsay. She offers just the right combination of a strong professional background and a sensitivity and understanding that comes from first-hand experience as the parent of a child who had a major health challenge.



Lindsay holds a Masters in Business Administration from Rowan University, as well as a Bachelors degree in Marketing and E-Commerce Technology from Rutgers University. She and her family live in New Jersey, just outside of Philadelphia.

Lindsay assumed her new position on October 24, 2011, overseeing and coordinating the work of the BSF staff, including Shelley Bowen, Director of Family Services and Awareness, Lynda Sedefian, Executive Assistant, and Dr. Matt Toth, Science Director. Lindsay will serve on the Board of Directors as an *ex-officio* officer.



The Barth Syndrome Foundation Board of Directors is also very pleased to announce its newest Board member — **Dr. David Axelrod**. Dr. Axelrod is a Pediatric Cardiologist at the Lucile Packard Children's Hospital at Stanford, in Palo Alto, California. He graduated from Dartmouth College in 1998 and received his M.D. from Ohio State University College of Medicine in 2002. Dr. Axelrod completed his internship and residency in Pediatrics at the University of California in San Francisco and completed his fellowship training in cardiology (2009) and intensive care (2011) at Stanford.

Although Dr. Axelrod had heard of Barth syndrome earlier, he became more fully aware of Barth syndrome through meeting Shelley Bowen at a medical conference in 2008, and shortly thereafter he helped diagnose and care for a Barth patient in California. His family's involvement with BSF dates back to 2002 when his then future wife served as a volunteer at the first official BSF Conference at Johns Hopkins in Baltimore.

Dr. Axelrod brings the insight of a practicing Pediatric Cardiologist, the personal and professional experience of one who has cared for a Barth syndrome patient and who holds an important position in a major California teaching hospital to the Board. His interest in and dedication to our mission has been clear from our first meeting and he is eager to help us improve awareness within the medical community and find more families on the West Coast.

Dr. Axelrod's term will run through April of 2015.

I know that I speak for the entire Board of Directors in welcoming Lindsay and David to our Board and into our small but strong and growing community. We are glad to have you both working with us!



(Photos courtesy of Lindsay Groff and Dr. David Axelrod - 2011)

Letter from the Executive Director

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



Lindsay with her daughter, Charlotte
(Photo courtesy of Lindsay Groff ~ 2011)

From the moment I read the job description, I was interested in learning more about the Barth Syndrome Foundation (BSF). Every line on that two-page document appeared carefully crafted with much thought put behind each word. I sensed this organization had accomplished great things, and I wanted to know more. The instant I started doing the background research in order to prepare for the interview process, I was captivated by an organization that has come such a long way in a short time and has done so much with so little. I read the articles in the newsletters detailing the amazing advancements made in research, new ideas for treatments, and heartfelt stories from families all over the world. I cheered while reading about the achievements, and I cried with the stories about those who have suffered.

When I received the offer to join the team, I felt honored and excited. I feel that my background in nonprofit management, coupled with my energy

and enthusiasm, will serve BSF well. I am looking forward to using my skills in management, marketing, and conference planning for all of the activities that support those affected by Barth syndrome. I believe that BSF is an excellent fit due to my passion for helping people cope with rare health issues.

My personal experience with a rare health condition came while pregnant with my daughter, Charlotte. She was diagnosed *in utero* with a birth defect called giant omphalocele, whereby the abdominal organs protrude into the base of the umbilical cord. Fortunately, she has made excellent progress and is now an active toddler. Through her, I learned about the isolation families often face, the feelings of being overwhelmed with medical and emotional issues, and the steep learning curve involved when handling medical equipment, surgical interventions, and complicated medication schedules. Having a child who has overcome a complex medical issue has changed my life forever. All of my experiences, both professionally and personally, led me straight to BSF.

In my first few weeks as Executive Director, I have had opportunities to speak to each member of the Board, staff, affiliate leads, and a few of the families. I look forward to meeting more families as I continue my "listening tour" as well as at the Conference. I also read many of the posts on the Listserv. Hearing your stories and experiencing the sense of community, I feel instantly included and connected.

Each of you plays an integral role in the prosperity of the Barth Syndrome Foundation. Generous donors provide the means to conduct research, raise awareness, and support programs. Tireless volunteers give selflessly to help manage an increasingly sophisticated organization. Scientists push for answers, dedicated to paving the way for the next breakthrough. Doctors advocate for their patients, looking at them as people and not solely as diagnoses. Together, this community offers hope, compassion and a common goal — to find a cure for the disorder.

Thank you for inviting me to be a part of this journey.

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Great News about Barth Syndrome Individuals' Longevity

(Cont'd from page 1)

These incredibly positive trends are the result of several factors. Part of the improvement is the consequence of more advanced medical knowledge and practices. Even though there is no treatment specifically for BTHS yet, more (though still not enough) is known about the various components of the disorder and the complexities of their interactions. Furthermore, medicine in general has advanced, and those who have BTHS are the beneficiaries of progress that is being made in a number of fields, such as cardiology and hematology, and in the medications that are being used to treat diseases in these areas.

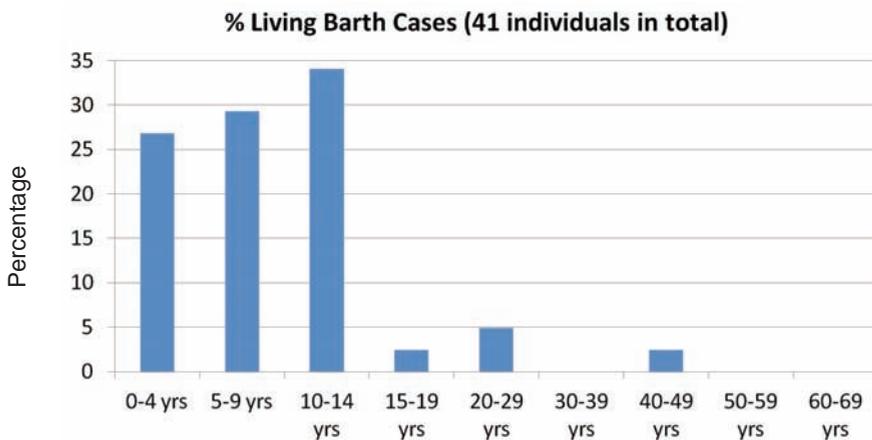
Additionally, more individuals are being properly diagnosed with BTHS. This has resulted from increased awareness of the disorder, both as a direct result of efforts that BSF and our affiliates have made, but also from the increased number of medical and scientific publications about the syndrome and the science that underlies its clinical manifestations. Actually,

BSF and our affiliates can take some credit for this as well, since now fully 60 peer-reviewed articles, published in a host of well-known professional journals, have credited BSF and/or our affiliates with support of the scientific work that is presented in them.

With all of this increased awareness, an interesting thing has happened — more adult men with BTHS are being discovered and finally are getting a correct diagnosis. Often this occurs after a young relative has received a diagnosis of Barth syndrome and it is realized that an older uncle or cousin, who had always been a bit “sickly” or who might have had a heart problem, also should be tested and is found to have BTHS.

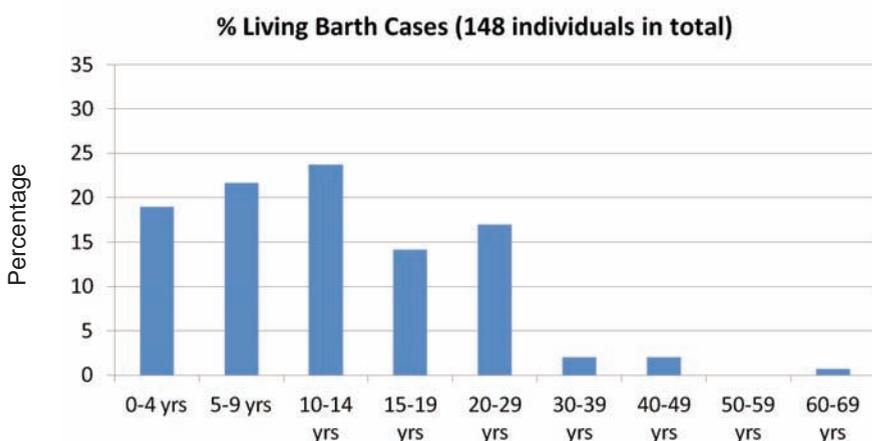
Whatever the cause, this improving trend in the longevity of those we know with Barth syndrome is indeed wonderful news for all of us. It is clear that we still have a long way to go before we have conquered the problems of BTHS. However, now we can show that those living with BTHS and their families can have real hope. Scientists can be assured that their work is resulting in a better understanding of this disorder, physicians can see the positive results of knowing better how to care for those with this syndrome, volunteers can prove that their investment of time and energy is paying off, and donors can be very confident that their funds are supporting efforts that are truly making a difference. Thank you, thank you, one and all. We can all rejoice together in this good news — at least momentarily, before we go back to work. Let’s all keep up the good work!

Year-End 2000



Graph #1

September 2011



Graph #2

(Source: Barth Syndrome Foundation, Inc.)

A Continuum of Collaboration

Barth Syndrome 2012 Scientific and Medical Sessions

(Cont'd from page 1)

The BSF has always eagerly sponsored the SciMed sessions of the Conference. There is a small group of BSF volunteers who work very hard in advance of the meeting to make sure that all the invitees feel welcome and are taken care of (herding cats comes to mind). For the 2002, 2006 and 2010 Conferences, the BSF was able to secure funding from the National Institutes of Health (NIH) to help defray a significant amount of the cost of these SciMed sessions. The BSF has applied to the NIH for funding the 2012 SciMed sessions as well. In 2010 and in other conference years, some of the SciMed participants were sponsored by the BSF affiliates in their own countries, while several invitees generously paid their own expenses to attend. Financial sponsorship of the SciMed sessions — from either the BSF directly, grants from the NIH, specific sponsorships by BSF affiliates, or the generosity of individual researchers — enables this unique and valuable Conference to advance the progress of and communication about Barth syndrome (BTSH) research.



A large part of the program for the SciMed sessions comes from the BSF Research Grant Recipients and other researchers doing work on BTSH. All of the recent BSF Research Grant Recipients for the previous two years are invited to come to the meeting and tell us about their work and what it means for the future. In addition to this resource, several members of the Scientific and Medical Advisory Board of the BSF either speak, chair specific sessions, or suggest speakers for the SciMed sessions. We try to include a keynote speaker at each Conference who will interest attendees from both the Family and the SciMed sessions. For 2012, we are very pleased to announce that the Director of the Office of Rare Disease Research (ORDR, a part of the NIH) and a great friend of the BSF, Stephen C. Groft, Pharm.D, will be our keynote speaker. Dr. Groft will speak on how ORDR and the NIH are expanding translational research activities in the growing area of rare disease research and how these efforts have become an important part of the continuing evolution of the NIH as the premier biomedical research funding organization in the world.

Poster Sessions

The Poster sessions have developed into another exciting part of the Conference. The Poster session allows both the SciMed and Family attendees to meet with the researchers in an informal setting — a setting conducive to education and relationship building. It is a rewarding development of these Conferences and of the Poster sessions in particular, to see friendships arise and evolve into professional collaborations. There are many fine examples where publications have arisen from just such personal encounters, and we expect 2012 to continue this important work. Often the Poster sessions are the primary area where the younger members of the BTSH research community are exposed not only to other researchers, but to BTSH individuals and their families. These interactions play a vital part in advancing and invigorating our research community for now and in the future.

New for 2012 is the offering of scholarship opportunities for young physicians to attend the SciMed sessions. We have been so pleased with the success of the poster stipend program to support the attendance of young scientific researchers that we wanted to institute a parallel program for young medical doctors. So, in response to the need to increase physician recruitment and involvement, this new program is intended to help encourage physicians and other healthcare providers, especially those early in their careers, to consider getting more involved with caring for BTSH individuals and with finding ways to increase the quality of their treatment. By combining motivated researchers with caring healthcare providers, the BSF seeks to make real advances in improving the future of BTSH individuals and their families.

Scholarships and Poster Stipends

BSF is pleased to be able to continue to offer stipends for those SciMed attendees who present posters. As described above, this year BSF also will offer a scholarship program for young healthcare professionals to attend the 2012 Conference. Both stipend programs are designed to defray the cost of attending the Conference and require applicants to fill out a form that describes their reasons for wanting to attend and their need for funds. All information and forms related to poster presenters and to the stipend programs are available on the BSF website at www.barthsyndrome.org.

(Photo courtesy of Cherie Schrader ~ BSF 2010 Conference)

Hooray — 2012 is a Conference Year!

By Sue Wilkins, Board Member, Barth Syndrome Foundation

I am so excited that 2012 is another BSF Conference year! The Conference is such an exhilarating, amazing, “there really are no words to describe” kind of event --- that NEVER disappoints. It’s one of those weeks that my family and I look forward to, and it flies by. Then it’s always so hard to say goodbye to everyone and wait two more years to see our Barth family again. And, hooray!!! It’s almost time; 2012 is a Conference year!

We will be at an historic, incredible location — the Pink Palace — otherwise known as the famous Don CeSar Hotel, right on the beach in St. Pete Beach, Florida. I understand that the hotel has fun activities for kids and that there are things to do outside of the hotel. But MOST importantly, we will all be together again.

At the risk of sounding like the BSF “matron” that I am....when Shelley Bowen, Anna Dunn and I (with the help of our own, fabulous Dr. Richard Kelley) planned the very first meeting of what became the start of our Barth organization in 2000, we did so on a wing and a prayer. We were three driven mothers determined to find more families living with Barth syndrome (BTHS). We had no earthly idea that it would be the beginning of finding all of you! We all are on this journey together to help our sons — your son and mine.

As we often say, especially when a new family joins us, this isn’t a group any of us would choose to be a part of, but we are so grateful that we are all in this together and have each other. Each of YOU brings more information, more experience, more knowledge about Barth syndrome -- whether your child is an infant or 20 years old. YOU are the expert about how BTHS affects your son and your family. Each of YOU is an incredibly critical link in the chain of discovery to help each of our sons. I don’t know about you, but each of YOU is a part of my family like no other. YOU get it. YOU instantly understand. YOU are the connection to living with BTHS that I can’t get anywhere else in the world. So...that’s why I’m so excited that, in a few short months, we’ll be together for another week! It really is like a “shot in the arm”... a BTHS family booster that keeps us going for another two years. And I for one, cannot wait!



Who says our BTHS boys can't perform like Beethoven
(Photo courtesy of Cherie Schrader ~ BSF 2010 Conference)

Based on the feedback you have given in evaluations and surveys, we are in the process of designing the 2012 Conference agenda and schedule. Details will be available soon on the web and listserv, and we can assure you it will be another educational and fun meeting!

I know that many of you are making plans to attend, having fundraisers to help defray the costs and arranging for time off work. For those of us affected by and living with BTHS in our families, I cannot imagine a more important week to be a part of... and it comes by only once every two years. Not only will we have the opportunity to sit with the world’s experts on BTHS, to ask questions, to hear the latest research results, to visit with the scientists, researchers and doctors, but ALSO to have a week to talk face to face with the world’s experts in living with a child with Barth — each other — and to give our kids --- affected boys and their siblings --- ONE week in two years to be together. As one new mom at the last conference said, “My son is normal here.”

I’ll end as I’ve started — my family and I cannot wait to see you and your family in a few short months on the beach at our 2012 BSF Conference!!!

Family Sessions at BSF's 2010 Conference
(Photos courtesy of Cherie Schrader ~ 2010)



6th International Barth Syndrome Scientific, Medical & Family Conference

June 25-30, 2012
St. Pete Beach, Florida

MAKE YOUR RESERVATIONS NOW!

A dedicated booking website has been created for this event so you will be able to make your hotel reservations on-line. To reserve your hotel room at the Don CeSar Hotel via the internet, please click the following link: <http://www.loewshotels.com/en/Don-CeSar-Beach-Resort/GroupPages/BarthSyndromeFoundation>.

To reserve your room by telephone, please call the Don CeSar Hotel Travel Planner (1-800-282-1116, or for callers outside the United States and Canada, dial International +1-727-360-1881, and reference "Barth Syndrome Conference" to guarantee the reduced rate (US \$125 per night). Beach House Suites are also available to the Group for US \$132 per night. These one bedroom suites include a full kitchen and are located approximately one-half mile north of the Hotel.

In addition to making your hotel reservation, you will need to register for this Conference with the Barth Syndrome Foundation. The registration form will soon be available on-line at www.barthsyndrome.org. For assistance or further information, please contact bsfinfo@barthsyndrome.org.

	Science & Medicine	Family
Monday, June 25, 2012		Registration & Welcome Reception
Tuesday & Wednesday June 26-27, 2012	Patient Discussions <i>(by invitation)</i>	Discussions with BTHS experts and Research Studies (formerly called "Clinics")
Wednesday Evening June 27, 2012	Physician/Scientist Registration and Informal Reception	
Thursday & Friday June 28-29, 2012	Scientific-Medical Sessions Poster Session (Thursday)	Family/BTHS Individual/Sibling Sessions
Saturday June 30, 2012	SMAB Meeting <i>(by invitation)</i> Closing Ceremony	Breakouts Closing Ceremony

Poster Stipends

As in previous years, a poster session is planned for BSF's 2012 International Conference. The poster session is an important part of the Conference, and the poster presenters are encouraged to apply for a stipend (up to US \$800) to help defray the cost of their attendance. Program and application information will soon be available at www.barthsyndrome.org.

Scholarship Program

New for 2012, the Barth Syndrome Foundation has a limited number of travel scholarships (up to US \$800 each) for qualifying physicians, clinical residents/fellows/students, nurses, and other allied health professionals to help defray the cost of attending the BSF's 2012 International Conference. This new program is designed to encourage medical practitioners to increase their knowledge about and improve their care of Barth syndrome individuals. Program and application information will be available soon at www.barthsyndrome.org.



Keynote Speaker

BSF is very pleased to announce that the Director of the Office of Rare Disease Research (ORDR), Stephen C. Groft, Pharm.D, will be the keynote speaker at BSF's 2012 Conference. Dr. Groft has been one of the most enthusiastic supporters of the BSF, and his department has been very generous in financially supporting our biennial conferences. The ORDR reports to the Director of the NIH, Dr. Francis Collins, whom you may know from his involvement with the acclaimed human genome project. The BSF is greatly appreciative of the NIH, and of the ORDR in particular, for their help and advice over the years. We look forward to hearing Dr. Groft's ideas about the future of rare disease research. *(Photo courtesy of Dr. Groft ~ 2011)*

Science Corner

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



Photo courtesy of Dr. Toth ~ 2011

One of the recent successes of Barth syndrome research that has been commented on by other scientists is the development of the mouse model. From the earliest stages of the BSF, the development of a mouse model of Barth syndrome (BTHS) was an important goal for the organization. Today, the *tafazzin* knockdown mouse model of BTHS is being studied in nine laboratories in the US, Canada, and Europe. In addition, this mouse line has recently been made available at the Jackson Laboratories (<http://jaxmice.jax.org/strain/014648.html>) to any interested researcher. Jackson Laboratories is the most famous research institution devoted to the use of the mouse as a model system to help understand human disease, and this prestigious institution doubles as a mouse breeding and production facility that supplies many types of mouse strains to researchers world-wide. The BSF hopes that by continuing to provide this mouse model to any researcher anywhere in the world, there will be an increase in work on and knowledge about Barth syndrome and other similar human diseases.

The tenth cycle of the BSF Research Grant Program started on October 31st of this year. To date, the BSF has given out 54 awards totaling over US \$2.0 million to 34 individuals and directly yielding 60 publications. There have been over 160 scientific/medical publications about or directly related to Barth syndrome, with over 130 appearing since 2002 — the year of our first BSF Research Grant cycle. I believe that a good portion of this publication increase is directly due to the BSF Research Grant Program. More significantly, the level of funding for BTHS research provided by the NIH and other organizations has grown substantially over the years (see figure below). The fact that other organizations have committed, and continue to commit, significant funding to BTHS research confirms and amplifies the value of the work that the BSF has nurtured from its very beginning. This year, one of the researchers using the mouse model of Barth syndrome, Dr. Zaza Khuchua, was awarded a 4-year NIH grant to continue his important studies of this model system. In addition, on May 31st of this year, Drs. Zaza Khuchua and Barry Byrne gave a webinar which described their results of the mouse model of BTHS.

Another important development was a recent publication concerning the clinical characteristics of BTHS individuals from data collected at the 2008 BSF International Conference. (Spencer CT, Byrne BJ, Bryant RM, Margossian R, Maisenbacher M, Breitenger P, Benni PB, Redfearn S, Marcus E, Cade WT. **Impaired Cardiac Reserve and Severely Diminished Skeletal Muscle Oxygen Utilization Mediate Exercise Intolerance in Barth Syndrome.** Am J Physiol Heart Circ Physiol. 2011 Nov;301(5):H2122-9. Epub 2011 Aug 26. This publication provides a starting point for comparative analysis with other mitochondrial diseases and shows some of the clinical problems that are faced by these individuals. The parent-practical outcome of this paper is that Barth syndrome individuals have a well-defined physiological impairment that causes them to fatigue very easily even with what would be considered reasonable exertion. It also provides scientific evidence to help justify requests that appropriate measures be taken to lessen the difficulties Barth syndrome individuals encounter when attending school, performing work, etc. We hope that this clinical paper and others will attract more physicians and scientists to be interested in our cause.

All Barth Syndrome Grants

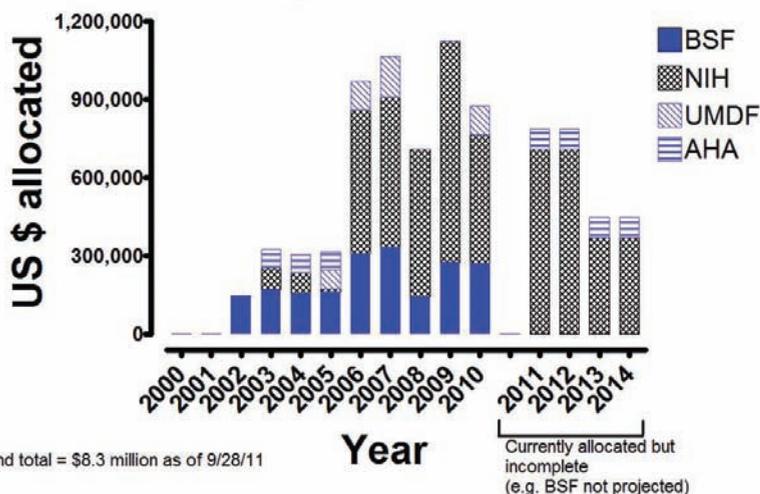


Figure 1. Graph of funding committed for Barth syndrome research.

BSF: Barth Syndrome Foundation Research Grant Program

NIH: National Institutes of Health

UMDF: United Mitochondrial Disease Foundation

AHA: American Heart Association

Amounts for 2011 and beyond correspond to what is allocated so far, but this amount is expected to increase given the previous history.

Sensory-Based Issues in Children and Adolescents with Barth Syndrome

By Roxanna Bendixen PhD, OTR/L, Consuelo Kreider MHS, OTR/L, and Stacey Reynolds, PhD, OTR/L; Department of Occupational Therapy, University of Florida, Gainesville, FL

“Salt. I put salt in my vinegar.... I’ll take vinegar, put salt in it, take ketchup put it in the vinegar, put salt and dip my fries in it.”
~ BTHS Individual

BACKGROUND: The ability to register and process sensory information (taste, smell, sound, touch, sight, movement) from the environment is believed to be one of the earliest tasks the human nervous system is capable of, forming the foundation for many higher level cognitive, motor, and social-emotional abilities. When sensory information is not integrated or processed appropriately, atypical behavioral responses and functional skill deficits may be seen. In some genetic conditions (e.g., Fragile-X syndrome and Angelman’s syndrome), atypical responses have been reported across sensory modalities (e.g., tactile, auditory, gustatory), with varying severity and consistency across the lifespan. Both sensory related *under-responsiveness* (i.e., failure to notice or respond to stimuli) and *over-responsiveness* (i.e., exaggerated responses to or avoidance of stimuli) have been documented, as well as behaviors associated with *sensation seeking/craving*. These behaviors may be identified very early in life, and problematic sensory behaviors may be modified with certain types of intervention such as sensory-based occupational therapy.

Barth Syndrome Study

At the 5th International Barth Syndrome Conference held in July 2010 in Orlando, Florida, three researchers from the University of Florida conducted the first scientific investigation of sensory-based behaviors in boys with Barth syndrome. An overview of the study is presented here while a full version of the manuscript is currently being considered for publication. The purpose of the study was to examine the prevalence of atypical sensory behaviors in boys with Barth syndrome and to explore if certain behaviors were present that may be useful for early diagnosis and treatment. Families attending the Conference participated in the study by completing interviews and questionnaires. Data were included from a total of 21 families with boys ranging from 3-17 years of age.

Based on results from the interviews and the Sensory Profile questionnaires, we found that sensory issues related to feeding and eating were ubiquitous in boys with Barth syndrome, with some behaviors such as strong gag reflex identifiable early in development. Specifically, boys with Barth syndrome had a very strong preference for salty, cheesy, and spicy foods, while having an overall restricted repertoire of foods they would eat (i.e., picky eaters). Boys identified as picky eaters often had strong taste, smell or texture aversions, with parents reporting that their child would not eat bland, lumpy, mushy, or slimy foods. Some families also noted that their children were sensitive to the temperature of the food/liquid, and items that were too hot or too cold were refused. Food issues were noted to impact on family function and often required adaptations to daily routines. Families reported buying certain preferred foods or altering foods (e.g., by adding salt) to meet the specific preferences of their child with Barth syndrome. Concerns about overall health and growth were raised by some families due to their child’s dietary restrictions.

Auditory differences, such as sensitivity to loud or unfamiliar noises and difficulty ignoring background noise, also emerged as sensory-related behaviors affecting performance and participation in boys with Barth syndrome. Some families/children reported difficulty concentrating in school or doing homework when there were competing noises in the environment. Other families noted that their child was bothered by certain sounds or could not handle the sounds at birthday parties or at the mall. Unlike feeding issues, however, these auditory behaviors were not described by parents as being problematic during early childhood (before the age of 3) and appeared to become more evident as the demands of school or social expectations (e.g. hanging out with a large group of friends) increased.

Overall, this study provides an initial step in describing and documenting sensory behaviors in children with Barth syndrome. The ability to process sensory information, and the impact atypical sensory behaviors have on function, should be further examined. Food-related sensory behaviors associated with taste, smell and texture sensitivity may be particularly important to consider in light of nutritional concerns expressed by many parents. Additionally, the possibility that auditory processing difficulties may be present in younger children with Barth syndrome should be further pursued prior to the children’s engagement in school and more complex social environments. While more work needs to be done, we hope that this research will help families better understand and identify sensory-based issues in their child and seek treatments which may minimize the impact of these behaviors on function and participation.

For further information, please contact Stacey Reynolds; Box 100164; University of Florida, Gainesville, FL 32610.
E-mail: Reynoldsse3@phhp.ufl.edu.

Awareness of Barth Syndrome is Growing Exponentially

There has been a significant increase in Barth syndrome (BTHS) related peer-reviewed journal articles published. To date, there have been a total of **60** articles 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.

1. Saini-Chohan HK, Mitchell RW, Vaz FM, Zelinski T, Hatch GM. **Delineating the role of alterations in lipid metabolism to the pathogenesis of inherited skeletal and cardiac muscle disorders.** *J Lipid Res.* 2011 Nov 7. [Epub ahead of print]†
2. Makaryan V, Kulik W, Vaz FM, Allen C, Dror Y, Dale DC, Aprikyan AA. **The cellular and molecular mechanisms for neutropenia in Barth syndrome.** *Eur J Haematol.* 2011 Oct 24. doi: 10.1111/j.1600-0609.2011.01725.x. [Epub ahead of print]
3. Claypool SM, Koehler CM. **The complexity of cardiolipin in health and disease.** *Trends Biochem Sci.* 2011 Oct 17. [Epub ahead of print]
4. El-Hafidi M, Meschini MC, Rizza T, Santorelli FM, Bertini E, Carozzo R, Vázquez-Memije ME. **Cardiolipin content in mitochondria from cultured skin fibroblasts harboring mutations in the mitochondrial ATP6 gene.** *J Bioenerg Biomembr.* 2011 Oct 13. [Epub ahead of print]
5. Momoi N, Chang B, Takeda I, Aoyagi Y, Endo K, Ichida F. **Differing clinical courses and outcomes in two siblings with Barth syndrome and left ventricular noncompaction.** *Eur J Pediatr.* 2011 Oct 7. [Epub ahead of print]
6. Wahjudi PN, Yee J, Martinez SR, Zhang J, Teitell M, Nikolaenko L, Swerdlow R, Wang C, Lee WN. **Turn-over of non-essential fatty acid in cardiolipin in rat heart.** *J Lipid Res.* 2011 Sep 27. [Epub ahead of print]
7. Takeda A, Sudo A, Yamada M, Yamazawa H, Izumi G, Nishino I, Ariga T. **Eponym : Barth syndrome.** *Eur J Pediatr.* 2011 Sep 23. [Epub ahead of print]
8. Takeda A, Sudo A, Yamada M, Yamazawa H, Izumi G, Nishino I, Ariga T. **Barth syndrome diagnosed in the subclinical stage of heart failure based on the presence of lipid storage myopathy and isolated noncompaction of the ventricular myocardium.** *Eur J Pediatr.* 2011 Sep 20. [Epub ahead of print]
9. Spencer CT, Byrne BJ, Bryant RM, Margossian R, Maisenbacher M, Breitenger P, Benni PB, Redfearn S, Marcus E, Cade WT. **Impaired cardiac reserve and severely diminished skeletal muscle oxygen utilization mediate exercise intolerance in Barth syndrome.** *Am J Physiol Heart Circ Physiol.* 2011 Nov;301(5):H2122-9. Epub 2011 Aug 26.*▼
10. Wortmann SB, Morava E. **3-methylglutaconic aciduria type IV: a syndrome with an evolving phenotype.** *Clin Dysmorphol.* 2011 Jul;20(3):168-9. No abstract available.
11. Saini-Chohan HK, Dakshinamurti S, Taylor WA, Shen GX, Murphy R, Sparagna GC, Hatch GM. **Persistent pulmonary hypertension results in reduced tetralinoleoyl-cardiolipin and Complex II + III activity in neonatal pig heart.** *Am J Physiol Heart Circ Physiol.* 2011 Aug 12. [Epub ahead of print]*▼
12. Taylor M, Slavov D, Salcedo E, Zhu X, Ferguson D, Jirikowic J, Di Lenarda A, Sinagra G, MD, Mestroni L. **Tafazzin gene mutations are uncommon causes of dilated cardiomyopathy in adults.** *Cardiogenetics.*2011.e4 | Published: 2011-07-05.*▼
13. Oechslin E, Jenni R. **Left ventricular non-compaction revisited: a distinct phenotype with genetic heterogeneity?** *Eur Heart J.* 2011 Jun;32(12):1446-56. Epub 2011 Jan 31.
14. Bockeria LA, Berishvili D, Baryshnikova I. **Re: ventricular non-compaction in children: clinical characteristics and course.** *Interact Cardiovasc Thorac Surg.* 2011 Mar;12(3):373.
15. Jacoby D, McKenna WJ. **Genetics of inherited cardiomyopathy.** *Eur Heart J.* 2011 Aug 2. [Epub ahead of print]
16. Paradies G, Petrosillo G, Paradies V, Reiter RJ, Ruggiero FM. **Melatonin, cardiolipin and mitochondrial bioenergetics in health and disease.** *J Pineal Res.* 2010 May;48(4):297-310. Review.
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18. Cavallo A, Gnoni A, Conte E, Siculella L, Zanotti F, Papa S, Gnoni GV. **3,5-Diiodo-L-Thyronine increases F(o)F (1)-ATP synthase activity and cardiolipin level in liver mitochondria of hypothyroid rats.** *J Bioenerg Biomembr.* 2011 Jul 8. [Epub ahead of print]
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20. Swallow H, Kirby DM, Blakely EL, Mitchell A, Salemi R, Sugiana C, Compton AG, Tucker EJ, Ke BX, Lamont PJ, Turnbull DM, McFarland R, Taylor RW, Thorburn DR. **Respiratory chain complex I deficiency caused by mitochondrial DNA mutations.** *Eur J Hum Genet.* 2011 Jul;19(7):769-75. doi: 10.1038/ejhg.2011.18. Epub 2011 Mar 2.
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22. Christie DA, Lemke CD, Elias IM, Chau LA, Kirchoff MG, Li B, Ball EH, Dunn SD, Hatch GM, Madrenas J. **Stomatin-like protein 2 binds cardiolipin and regulates mitochondrial biogenesis and function.** *Mol Cell Biol.* 2011 Jul 11. [Epub ahead of print]
23. Dale DC. **Editorial: Serine proteases, serpins, and neutropenia.** *J Leukoc Biol.* 2011 Jul;90(1):3-4. No abstract available.
24. Xiao J, Engel JL, Zhang J, Chen MJ, Manning G, Dixon JE. **Structural and functional analysis of PTPMT1, a phosphatase required for cardiolipin synthesis.** *Proc Natl Acad Sci U S A.* 2011 Jul 5. [Epub ahead of print]
25. Sandler Y, Baumann S. **Characterization of urine metabolites in Barth syndrome patients employing a non-targeted GC/MS screening approach.** 59th ASMS Conference on Mass Spectrometry, June 5 - 9, 2011, Denver, Colorado. (Poster)
26. Wajner M, Goodman SI. **Disruption of mitochondrial homeostasis in organic acidurias: insights from human and animal studies.** *J Bioenerg Biomembr.* 2011 Feb;43(1):31-8.

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)

NIH Director's Transformative Research Awards (RO1)

Funding Opportunity Announcement (FOA) Number RFA-RM-11-006

Open Date: December 12, 2011

Letter of Intent Due Date: December 12, 2011

Application Due Date(s): January 12, 2012

Expiration Date: January 13, 2012

<http://grants.nih.gov/grants/guide/rfa-files/RFA-RM-11-006.htm>

Purpose: The goal of the NIH Director's Transformative Research Awards initiative is to provide support for collaborative investigative teams or individual scientists who propose transformative research projects, which, if successful, would have a major impact in a broad area of biomedical or behavioral research. To be considered transformative, projects must have the potential to create or overturn fundamental scientific paradigms through the use of novel approaches or to lead to major improvements in health through the development of highly innovative therapies, diagnostic tools, or preventive strategies. Consistent with this focus, applications supported under the Transformative Research Awards initiative will reflect ideas substantially different from mainstream concepts.

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.

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&oppld=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.

Pilot and Feasibility Clinical Research Grants in Diabetes, Endocrine and Metabolic Diseases (R21)

Program Announcement (PA) Number: PA-09-133

Opening Date: May 16, 2009

Letters of Intent Receipt Date(s): N/A

Application Receipt/Submission Date(s): Multiple dates

Expiration Date: May 8, 2012

<http://grants.nih.gov/grants/guide/pa-files/PA-09-133.html>

Purpose: This FOA, issued by National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) and the Office of Dietary Supplements (ODS) of the National Institutes of Health, encourages exploratory/developmental clinical research related to the prevention or treatment of diabetes, obesity and endocrine and genetic metabolic diseases. The Pilot and Feasibility Clinical Research Grants Program is designed to allow initiation of exploratory, short-term clinical studies, so that new ideas may be investigated without stringent requirements for preliminary data. The short-term studies should focus on research questions that are likely to have high clinical impact. They can include testing a new prevention strategy, a new intervention, or unique combinations of therapies. A high priority is the use of such studies to help stimulate the translation of promising research developments from the laboratory into clinical practice in diabetes, endocrine diseases and genetic metabolic diseases, including cystic fibrosis.

(Cont'd on page 12)

Funding Opportunities Relevant to Barth Syndrome Research

(Cont'd from page 11)

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)

UMDF Clinical Research Fellowship Training Award

Application Due Date: February 15, 2012

Statement of Intent Receipt Date: July 1, 2012

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.

Barth Syndrome Foundation Family Outreaches



Families from Nebraska, Iowa, and Indiana gather for BSF's Great Plains Family Outreach



A few of our Barth boys and young men at BSF's Great Plains Family Outreach



Families and friends from Florida, Georgia, and Texas gather for BSF's Southeast Family Outreach (ALL photos courtesy of BSF ~ 2011)

Great Plains Family Outreach

By John Wilkins, Board Member, BSF

The first-ever Great Plains Outreach was held June 10–12, 2011 in Lincoln, Nebraska — it was a big success! The Barth Syndrome Foundation encourages families to plan regional outreaches during the off-year of the BSF International Conference. I had been eager to host a regional outreach in Nebraska for a long time, and this was the year it all came together.

Our event allowed families from Nebraska, Iowa, and Indiana, in the United States, as well as Saskatchewan, Canada to spend time together. Families gathered on Friday night for a cook-out in my parents' back yard. Most of the Barth boys were around the same age. They played with a wooden train set I loved when I was little, as well as with bubbles and other outside toys as everyone got to know each other.

Saturday was a very busy day. We spent the morning and had lunch at the Lincoln Children's Zoo, where we saw many different kinds of animals. The little ones got to ride ponies, and all of us rode the train that encircled the zoo. After naptime, and swimming at the hotel pool, our group gathered at one of our city parks for pizza and lots of playing on the huge array of playground equipment. Sunday, several families spent time at my parents house relaxing and talking.

What I enjoyed most and feel was the most important thing for me was the fellowship with the other families. It was such a joy to be able to spend time with others who know and understand what Barth syndrome is after all these years of being alone. I especially enjoyed watching the younger boys play together and begin to form what will hopefully be lifelong friendships.

Southeast Family Outreach

By Shelley Bowen, Director, Family Services & Awareness, BSF

In June of 2011, several families gathered in Steinhatchee, Florida at the beautiful Steinhatchee Landings Resort. We wondered if we should move forward with so few families coming, but two years was simply too long a time span between conferences not to see one another.

The culture of friendship and devotion to one another within this community extends beyond the disease that initially brought us together. The commitment to the people within this community also extends beyond families who have been directly affected by the disease. That was evident during our weekend together. Six dedicated volunteers also came to the Landings to share time with the families they have come to know and care about over the years.

(Cont'd on page 14)



Julie, Angelo, and Shelley at BSF's Southeast Outreach

Southeast Family Outreach

(Cont'd from page 13)

As usual, we stayed up way too late and woke up way too early so that we could get the most of every precious moment together. We canoed down the Steinhatchee River; spotted wildlife and in some cases paddled like mad to avoid some of the wildlife we encountered during our excursion.

Michael Bowen and Sam were nowhere to be seen for about 30 minutes after the third canoe arrived at our destination point. A local fisherman was recruited by a BSF volunteer and designer of the BSF Blue Lemonade stand (Jim Zurbrick) to take me up river to search for our wayward paddlers. We found them taking a break on the bank of the river. Michael was to be towed back. Sam was thrilled because he caught a fish! The adults' arms were throbbing after four hours of paddling. All the children were happy to be back at the Landings for an afternoon swim.



Some of our Barth boys, young men (and one sibling!) at BSF's Midwest Family Outreach

The petting zoo was a big hit with the children. I don't think the koi in the koi pond were intended to be a part of the petting zoo, but that didn't stop the kids and the men from sticking their hands in the pond. As always, it was sad to say goodbye at the end of the weekend. We were all surprised to learn that Angelo Florez, Michelle Garcia, and her son, Matthew had taken a three-hour side trip to Orlando to go to Walt Disney World when we saw pictures of them at the park.

Midwest Family Outreach

By Tiffini Allen, BSF Volunteer

The weekend of July 22-24, 2011 was a scorcher in Indianapolis, as heat warnings adorned television sets and radios. This didn't stop Devin, Henry, Wyatt, Noah, John, Dylan and Peyton, and their family members from traveling a total of 1,557 miles to join together for the Midwest Barth Syndrome Outreach.

Friday, friends were reunited and new friendships were created. We cooked out, played baseball, rode tricycles, the kids chased each other around the house and the yard. Red faces were aplenty as the evening weather didn't show any signs of a cool down. The laughter never ended!

Saturday morning we all met at the Indianapolis Children's Museum, as dinosaurs, trains and Dora the Explorer awaited our arrival. Later that evening, we were joined by Henry's favorite hematologist, Dr. Grzegorz, as he joined us for pizza and a very special birthday celebration. Seven candles were atop a mandarine orange cake and exciting gifts surrounded. Happy Birthday was sung by all as we celebrated Devin's 7th birthday. Once the singing was finished and the cake was eaten, baseball was again played, tricycles were rode, and lots of kids with red faces continued to run around the house and yard. Non-stop laughter was flowing through the windows.

Heat warnings were finally lifted on Sunday as we all met at the public pool to swim, relax, share more laughs, and catch up before the outreach came to an end. We couldn't have asked for a more wonderful weekend.



(L-R) Dr. Grzegorz, Henry, and Tiffini at BSF's Midwest Family Outreach. *(All photos courtesy of BSF ~ 2011)*

My Story and Relationship with BSF

By Ashley Cade, St. Louis, MO

“...I watched and witnessed their hope, sacrifice, love, and care. When I looked at their son, my heart overflowed with joy and hope. I had an urge to want to fight for him, to stand up for him, to pray for him, to hope beyond all hope that he lives a full life and knows his family’s love. These were my emotions before I held him! Holding him for a few brief moments gave me a perspective that has stayed with me. It is a perspective that causes me to squeeze my girls a little tighter and pray more fervently for my friends with Barth syndrome.”



Ashley and Todd Cade with their daughters Lucy (L) and Emily (R)
(Photo courtesy of Ashley Cade ~ 2011)

My story and relationship with Barth syndrome (BTSH) is a little different from most. I am the wife of a medical researcher. This can make marriage complex when trying to understand what exactly my husband does all day. I do my best to be involved in a support role. However, I frequently need him to use a dry erase board to spell out, in layman’s terms, what he is researching. I love my husband with all my heart, and I love that he loves what he does for a living. He wants to make a difference, and I have always longed to make a difference alongside him.

Todd and I are also joyous parents to our daughters, Lucy, 4, and Emily, 22 months. Outside of God, family is the most important thing in my life. Every day with them is a gift. Every giggle and fight, every diaper blow out and potty success, every smile and tear; they are all precious gifts wrapped in brief moments. I know I take them for granted. But being alongside Todd and learning more about BTSH has given me a better perspective and helped me not take so much for granted.

First, it’s important to share how Todd (and later I) became involved with BTSH. It wasn’t planned. In fact, it was serendipitous. In 2006, Todd, was invited to dinner with Dr. Kevin Yarasheski, a close colleague at Washington University in St. Louis. Kevin had invited Dr. Barry Byrne to the medical school to give Metabolism rounds on gene therapy and how this relates to muscle disease; not necessarily BTSH. The evening before rounds, Todd, Dr. Byrne, and Dr. Yarasheski went to dinner. During the meal, Todd shared his experience with exercise testing and Dr. Byrne in turn shared information about BTSH.

When Todd came home from having dinner with Dr. Byrne, he had an excitement about him that said without words, “I think I can make a difference in boys with BTSH.” Evidently, Dr. Byrne had similar thoughts, as he invited Todd to the 2006 BSF

conference to assist in an exercise testing study with him and Dr. Carolyn Spencer a few months later. Todd’s enthusiasm was contagious, and I was intrigued. Once he explained the medical details of BTSH, my heart instantly opened and ached for the families. I wanted to meet them, love them, and understand the impact of what was being done to treat and cure BTSH.

My first opportunity was summer 2010 in Orlando, Florida. I accompanied Todd, with our girls, to the conference. I wanted to jump right in and know as much as possible. I was met with so many friendly faces and kind people. I met a few families, but I quickly started to feel like an outsider; not due to how I was treated, rather, how I felt on the inside. A voice in my head started whispering, “who are you to assume you can connect with families who are facing a hardship you could never begin to understand?” Honestly, I allowed it to shut me down and withdraw.

But then I met a family whose son was Emily’s age, 7 months at the time, and who had BTSH. At first I watched; then with little confidence remaining from the voice in my head, I introduced myself. I listened and was not in a hurry to talk (unusual for me). I watched and witnessed their hope, sacrifice, love, and care. When I looked at their son, my heart overflowed with joy and hope. I had an urge to want to fight for him, to stand up for him, to pray for him, to hope beyond all hope that he lives a full life and knows his family’s love. These were my emotions before I held him! Holding him for a few brief moments gave me a perspective that has stayed with me. It is a perspective that causes me to squeeze my girls a little tighter and pray more fervently for my friends with BTSH.

My hopes for the immediate future are that Todd be awarded an RO1 grant to further his research and that we raise local awareness in St. Louis about BTSH. We are delighted to have been selected to participate in the 2011 Winter Wonderland light display in Tilles Park. Visitors from far and wide come to this magnificent display of thousands of twinkling lights. As an organization that has been selected to participate, we are awarded a small financial gift and the opportunity to hand out information to vehicles that drive through the light display on our assigned evening. Many of our friends have already committed to volunteering on Thanksgiving night, our assigned evening! We are excited not only to connect with friends and family over BTSH but to bring awareness to our community. We believe this can be a spring board to establish an annual fundraiser in St. Louis and draw closer to the BTSH community. We will certainly be giving thanks for all the Barth families and their boys this turkey day!

My Strong Volunteer Connection with Barth Syndrome

By Bruce Justin (BJ) Develle, Social Worker, Florida



(R-L) BJ Develle together with his wife, Greta, and daughter, Georgia

In 1998, while away from home at college, I met the Bowens and was instantly welcomed as part of their family (home cooked meals, laundry done, comfortable bed, etc). In hanging out with them, I began to better understand Barth syndrome and witnessed their family's search for information, along with that of other families in a similar situation. When I had the opportunity to meet other parents at a meeting in Steinhatchee, FL, I felt instantly welcomed as one of their own. However, it wasn't until the first conference at Disney when I met the other affected young men that I cemented my connection with BSF. The young men and siblings I worked with during that conference amazed me with their maturity, understanding and perspective about Barth syndrome and how they were affected. That week, I met more BSF families from around the world and found myself being pulled in as

one of them. Given how close this community is, I felt honored to be greeted with so many open arms, hearts and emotions. From that point on, phone calls or emails from other states and countries became a welcome part of my life, as I kept in contact with those young men and their families. I heard more about their everyday lives, and they inquired about mine. We have shared excitement about successes and opportunities, anxiety about the unknown, and consoled each other when necessary.

After the conference, I placed objects and pictures in my office and at home as everyday reminders of this community. They provided encouragement for me to become more involved; spreading information about Barth syndrome and the Foundation, assisting with conference planning and programming, as well as utilizing my professional training and skills to provide guidance and assistance to individual families. Over the years, I have attended planning meetings, multiple conferences and even travelled internationally to work with this community. During this past year, I was approached by the Barth Syndrome Foundation of Canada on a professional level to interview their affected young men and provide guidance into the needs of their community. Following that, they invited me to come to Canada and meet with several of the young men for a discussion about addressing and overcoming relationship obstacles emerging from Barth syndrome. The meeting with the affected males went very well, and, of course, I loved getting to spend more time socially with all of them.

I look forward to more events and opportunities with BSF and its community, both professionally and personally, and I feel honored to be a part of this amazing family.



Editor's note: BJ currently works for the state of Florida's Agency for Health Care Administration interpreting policy and monitoring Substance Abuse and Mental Health providers. Previously, he provided case management and therapy services to children and specialized training to foster parents and professionals who would work with them. He has worked with both males and females with histories of physical and sexual abuse, brain injuries, mood disorders, drug exposure, suicidal and homicidal attempts and psychiatric residential placements, both in the community and within a group home he managed. BJ graduated from Florida State University with a Masters in Social Work in 2008, after earning Bachelors degrees in Child Development and Religion in 2002.

Greta and BJ with their daughter, Georgia, and husky, Loredo
(Photos courtesy of BJ Develle ~ 2011)

A HUGE Thank You to BSF Supporters

Together we can make a difference!

By Stephen McCurdy, Chairman, Barth Syndrome Foundation



JDRF & Barth Syndrome Foundation 6th Annual Golf Classic

Randy Buddemeyer and his partner at Grubb & Ellis held their 6th annual golf outing to support the Barth Syndrome Foundation and the Juvenile Diabetes Research Foundation. Despite the recession and challenges in the real estate world, Randy's friends came through once again and made a considerable contribution to BSF's programs. Oh, and by the way, they had another great time playing golf, bidding on auction items, and celebrating their exploits on the fairways and greens (and bunkers and traps!).

(Photo courtesy of Grubb & Ellis ~ 2011)



Henry and his buddies, Keegan and Colin, overlooking Wrigley Field *(Photo courtesy of Tiffini Allen ~ 2011)*

Hey Hey Henry Fundraiser

Henry's Mom, Tiffini, is a fund-raising and awareness powerhouse for BSF! Tiffini held the Chicago Cubs Hey Hey Henry Rooftop fundraiser where she rented out a rooftop overlooking Wrigley Park to watch a Cubs game and raise money for BSF... quickly followed by a Barth Walk along the canal in the Allen's hometown of Indianapolis. "The walk was along the canal where the hotel for the Indianapolis outreach is located. The canal is fantastic! We have met so many great friends through Henry's Barth syndrome appointments at Riley Children's Hospital, and several of them attended the Cubs game!" reported Tiffini. She has also arranged to have the BSF logo on participants T-shirts for several local walks and bike rides to increase awareness among the participants and fans, and her own Mom held a bowling fund-raising event for her friends and raised money for BSF as a part of the Hey Hey Henry fundraiser.



Slim and others
(Photo courtesy of Chris Hackett ~ 2011)

"SLIM"

Greg Holly has a brother-in-law with a great sense of humor, a passion for Barth syndrome, and a willingness to do almost anything to get attention! Greg reports on the May 7th Texas Rangers/New York Yankees baseball game: "Watch for me and my brother-in-law in the section behind home plate. He is the one who paints himself blue (as "SLIM") to promote BSF awareness at the Dallas Cowboy football games. Tonight, SLIM is going to the Rangers - his head will be painted red and he will be sportin' the BSF logo. He has challenged his FB friends who see him on TV to make donations to BSF. We lucked into some sweet tickets with the rich folks (not too far from where former Pres. Bush sits when he's there). Hopefully, we don't get "moved" before the game is over – LOL." Even the disappointed Yankees fans (Rangers won 7-5) had to admit that SLIM made a statement that night!

(Cont'd on page 18)

A HUGE Thank You to BSF Supporters

(Cont'd from page 17)



Baffa Beach Bash

Rosemary Baffa and her son, together with the Baffa clan know how to throw a party! (Did anyone doubt it?) On June 24th, the Great Hall at Immaculata College outside of Philadelphia, PA was filled with friends and relations enjoying the Baffa Beach Bash benefiting BSF. Great music, food and dancing were the order of the evening, all for a \$30 per person contribution to BSF at the door. The party was a smash hit and now the kids want this to be an annual event! How about it, Rosemary?

Supporters of Baffa's Beach Bash

(Photo courtesy of Rosemary Baffa ~ 2011)



Volleying for Barth

(Photo courtesy of Bryan Drake ~ 2011)

Co-ed Grass V-Ball Tournament

Bryan Drake, the father of a 4-year old son affected by Barth syndrome, decided to use his love of volleyball and his V-ball friends to raise money and awareness for Barth syndrome and BSF. He organized the first annual Barth Syndrome Co-ed Grass V-Ball Tournament which attracted 10 teams of four players each to Sunnyside Park in Kansas City, MO on September 17th. Bryan collected items from local business to raffle off, encouraged donations, and charged a \$30 minimum registration fee which the competitors were glad to contribute. He hoisted the BSF Banner and passed out BSF informational brochures, bracelets and newsletters. The event was a great success and we and Bryan's V-Ball competitors are hoping for a chance to repeat next year.



Members of Team Will and some of their loyal supporters

(Photo courtesy of Eliza McCurdy ~ 2011)

Lake Placid Ironman

Coach Gary Rodbell and Team Will fielded the largest Ironman team yet in Lake Placid, NY on July 24th! Gary, Ghent Lummis, Matt Karp, Laura Azar Kuhn, Heather Segal, Stefan Tungez, and Jaime Jofre plunged into the cold waters of Lake Placid at 7AM and all completed the 2.4 mile swim, 112 mile bike and 26.2 mile run well before the midnight cut-off time. Wearing their Barth Blue jerseys with the International logo on the back, they were easy to spot and made a real statement for us throughout the race. The team and their supporters once again raised money to support research and BSF programs for the Barth boys, young men, and families. This same team was then joined by the rest of Team Will for the Olympic length (a mere 32.1 mile) Jarden Triathlon in Rye New York on September 25th, again wearing their Barth Blue jerseys. Apparently, the skills and persistence required by fund-raising and Ironman Triathlons are very similar! Anyone who wants to join the US or French team is welcome!

(Cont'd on page 19)

Together we can make a difference!

(Cont'd from page 18)



Gene KingQueen Pageant

Many of you know Amanda Clark for her striking photos of the boys and their families that she takes at every Barth Conference and which appear in all of our BSF publications. Krista Vann, who works with Amanda, has also become a Barth devotee, and she organized a local children's beauty pageant as a fundraiser for BSF in Florida. The kids (and their Moms) had a wonderful time as you can see! We are delighted that Amanda and her team will be at our Conference again in 2012!

How adorable is this little guy modeling for BSF
(Photo courtesy of Krista Vann ~ 2011)



Winter Wonderland — Tilles Park, St. Louis

Ashley Cade is the wife of Dr. Todd Cade who has been leading research on exercise intolerance among Barth syndrome boys and young men (his article was recently published and sheds new light on the exercise challenges our boys and young men face and the accommodations they may require in school and at work. *(for full citation see page 10, #9)*). Ashley attended our 2010 Conference and has met many of the boys and their parents as they have traveled out to St. Louis for testing. She has become an avid supporter and member of the BSF community *(see Ashley's article on pg. 15)*. One of the traditional events held at holiday time in St. Louis is the Winter Wonderland in Tilles Park. From Thanksgiving through New Year, visitors are invited to drive or take a carriage ride through the festive displays and holiday lights in the park, and each year, a few charities

are selected to raise money and awareness among visitors to the park for a single night. Ashley took it upon herself to apply to St. Louis Parks and Recreation, and, because of her efforts, BSF was selected to hand out informational materials to everyone who visited the park on Thanksgiving evening, Nov. 24th! What a great way to spread awareness of Barth syndrome!



8th Annual Barth Syndrome Bowling Fundraiser

"Let's bowl!" seems to be the rallying cry of the Higgins Family! For the eighth year in a row, John and Liz Higgins lead their friends and family to the local lanes for an afternoon devoted to knocking down pins and building up the BSF bank account. After eight years, the crowd must be getting pretty good at bowling — they are certainly great at raising money and awareness for BSF, though by this point, everyone in Warwick, NY and the surrounding towns knows about the Higgins and Barth syndrome... they are making us all famous!

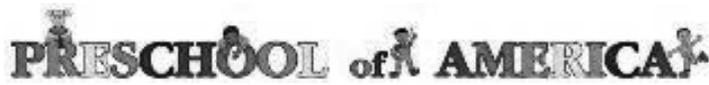
Family and friends come out to support BSF in Warwick, NY
(Photo courtesy of Higgins Family ~ 2011)

(Cont'd on page 20)

A HUGE Thank You to BSF Supporters

Together we can make a difference!

(Cont'd from page 19)



Preschool of America Special Art Project Day

For anyone who needs proof that the devotion of family and friends knows no geographic or language boundaries, look no further than to Raphaël. Raphaël lives in France and his

parents have started the newest Barth affiliate there. Friends of the family live in New York City and their story of Raphaël inspired the Preschool of America to encourage all of the preschoolers above the age of 2 to create personalized art projects in support of Raphaël. The result? The families of all these children now know about Barth syndrome and Raphaël received a host of messages and artwork wishing him and all Barth syndrome boys and young men well. Merci beaucoup Raphaël!!

Year-End Appeals

The McCurdy's are preparing to send out their annual year-end holiday letter asking their friends and family to support BSF. This is always an effective fund-raising effort simply because it is a personal appeal to people who know their son's story, the McCurdy family's devotion to BSF, and their determination to find a cure for Barth syndrome. Steve has any number of sample letters he has used in the past ten years and would be happy to share them with anyone who would like to follow suit. The McCurdys and the Wilkins, who also make good use of the US Mail service, find that their friends are actually looking for ways to be helpful and that contributions are willingly offered and gratefully received. Please join us in this inexpensive and effective way to raise funds for a cause near and dear to all of our hearts!

Donations Received In Honor Of or In Memory Of

BSF receives many donations **in honor of** special people. Over the last 18 months, donors have honored: Henry Allen; Laura Azar Kuhn; Kevin Baffa; Sarabeth Berman; Bly Brody; Andrew Buddemeyer; Thomas & Morlene Butera; Abram Drake; Jake Fairchild; William Floyd; Jack Higgins; Wallace Hopp; Jaime Jofre; Milosh Kalapasev; Matt Karp; Ghent Lummis; Ben Mann; Will McCurdy; Susan McCormack; Jack Moreland; Evan and Sarabeth Osnos Wedding; Gary Rodbell; Heather Segal; Ryan Sernel; Maryann Spatz; Stefan Tunguz; Nancy Wadel; Esther Pittack; John Wilkins; Muriel Wilkins; Sue Wilkins; Corey Wadel; Christopher Weller; Alex Wynia; and Greyson Zeitner.

Donations also have been received in memory of: Esther Anderson; Theodore Baffa; Michael Bowen, Jr.; MaryEllen Davis; Aden Edsel; Mavis Gourley; Norman Harris; Caleb Holly; Wallace Hopp; Sandra Moreland; Gloria Percival; Esther Pittack; Gertrude Poe; Allene Russell; Michael Telles, Jr.; Ben Thorpe; Evelyn Verona; and ALL of the Barth boys we have lost.

Barth Syndrome Foundation Receives GuideStar Exchange Seal as a Demonstration of Its Commitment to Transparency

The Barth Syndrome Foundation (BSF) is pleased to announce that the Foundation has received the GuideStar Exchange Seal, a leading symbol of transparency and accountability provided by GuideStar USA, Inc., the premier source of nonprofit information. The Seal demonstrates to BSF's support-base our deep commitment to nonprofit transparency and accountability.

We have worked hard to showcase our progress toward our mission and our long-held belief in being transparent about our work to our constituents. As a GuideStar Exchange member, we use their platform to share a wealth of up-to-date information about our work to our supporters and GuideStar's immense online audience of nonprofits, grantmakers, individual donors, and the media.

In order to be awarded the GuideStar Exchange Seal, BSF had to fill out every required field of our nonprofit report page on www.guidestar.org, including our annual report, photos, videos, etc.

BSF encourages you to look at our profile on GuideStar to see what we're all about. We are engaged in exciting initiatives, and we are thrilled to have another platform for communicating our advancement and progress.

Barth Syndrome Trust

Letter from the Chair

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

Hello everyone and a warm welcome to our new families. It was wonderful to see some of you at the last Bristol clinic and gathering in September this year. Our family thoroughly enjoyed the whole experience. Although the actual clinic was busy, it ran like clockwork, and we came away feeling reassured that Nick had been seen by everyone and had all the tests he needed, without being totally exhausted. In fact, he had most of the afternoon to rest and play with his friends, while we went to the hands-on training sessions and talks in the conference room. We adults had the opportunity to learn or revise our resuscitation skills; there was also detailed information about the home service for G-CSF, which consists of the supplies being regularly delivered to our homes, and a trainer who does home visits to train parents and children to do the injections. Lastly, there was a presentation about the role of nutrition in Barth syndrome, with special emphasis on amino acid supplementation as well as even a chance to taste arginine and cysteine in a variety of cocktails! (Note: they tasted better than we'd expected, especially when disguised in lime or blackcurrant juice.)

The Family Gathering at Avon Valley Country Park on 1st October coincided with a very welcome heatwave, and everyone relaxed and enjoyed the wonderful barbeque put on by Greg and Kerry-Ann Manton. There was a host of outdoor activities for the children, so fun was had by all. We particularly enjoyed Jack's culinary efforts to make meringue baskets for each of us. He did a great job of loading up each meringue with fruit and cream, with a little lick of the spoon he was using for the cream each time! Delicious!

DATE FOR YOUR DIARY

Barth Syndrome NHS Clinic

1st — 2nd March 2012

Bristol Royal Hospital for Children

Followed by a Family Gathering on Saturday 3rd March, venue TBD

Introducing New Trustees

The Trustees are delighted to welcome two new enthusiastic members to the Board.



Helen Coleman

My son is now 22 and had a heart transplant when he was 2 years old, (before we knew about Barth syndrome) and a renal transplant from his Dad when he was 13. He leads a pretty normal life now; working and driving. I have been married to Michael for 31 years and we also have a daughter, Ellie, who is now 17. I hope that by becoming a Trustee, I will be able to share with new and existing families the

experiences we have had over the years, and offer them hope and support. I bring to the Trust administrative skills gained from running our own business and current employment as a retail manager for Champneys Forest Mere.



Suzy Green

I am Mum to Mitchell, age 5, who has Barth syndrome. I was a social worker before Mitch was taken ill, and I now keep my hand in by doing 12 hours a week. As the mother of a young son, I can speak for younger families on the Board. I love fundraising and will tackle almost anything. I hope my enthusiasm for fundraising can make a difference to BST.

Barth Syndrome Trust

Letter from the Chair

(Cont'd from page 21)

New Neuropsychological Testing for UK Community

Barth Syndrome Trust (BST) funds Assistant Psychologist position for one day per week.

In recent clinics, parents have highlighted their concerns that many boys with Barth syndrome seem to struggle with attention. BST is working closely with Dr. Vanessa Garratt (Psychologist at Bristol Royal Hospital for Children) who will be leading a project to investigate this issue. From 2012, all boys and men who are interested in participating will undergo full testing at their homes or schools, in particular to identify whether there are any areas which could require intervention. This is not a traditional research project where participants do not usually get individual feedback. A full report of all findings with specific recommendations will be given to each family and the school. Once the results are collated and any trends identified, a UK-specific Educational Brochure will be created (similar to the existing BSF Educational Booklet), and these will be freely available.

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

Next BST Workshop for all Volunteers — We Need You!

Find out more about how you can help us achieve our mission. Learn about our programmes: raising awareness, publications, fundraising, administration and family services. There will also be a special focus group for families about education. Results from this focus group will be used in the Education Brochures we're creating with Dr. Garratt and Debbie Riddiford.

Venue: St Mary's Church Hall, Overton, Hampshire

Date: Saturday, 21st January 2012

Time: 10am – 5pm

Lunch provided
Help with travel costs



Suzy Green hands fundraising cheques to BST Treasurer, Gemma Wilks. (Photo courtesy of BSTrust ~ 2011)



2011 Trustees' meeting. (Photo courtesy of BSTrust ~ 2011)

(Cont'd from page 22)

News from other groups with whom we work closely...

From the Cardiomyopathy Association

In our last newsletter, we featured young Joe. By way of update, he recently received a new heart after being on a Berlin Heart for the longest time ever for a child at Great Ormond Street Hospital. The Berlin heart is an assistive device which is usually used as a bridge to a transplant.

Matthew Green, 40, is the first person in the UK to receive a new totally artificial heart (SynCardia temporary Total Artificial Heart) while waiting for a heart transplant. It is powered by a portable power pack that can be carried in a back pack, and, although there is a risk of infection and it cannot be used long term, it does mean that he can wait for a transplant in his own home.

Advances like these have been life-savers for people who are awaiting a heart transplant, especially these days when donors are so few. If you would like to become an organ donor, please visit www.organdonation.nhs.uk for further details and registration or amendments.

Disability Living Allowance (DLA) and new Welfare Reform Bill

Currently, a child's Disability Living Allowance (DLA) is stopped after 84 days in hospital, and subsequently the parents' Carer's Allowance is suspended. Contact a Family, The Children's Trust and many MPs are calling for these regulations to be scrapped. Further information is available from Contact a Family's website <http://www.cafamily.org.uk/campaigns/index.html> and <http://www.edms.org.uk/2010-11/1520.htm>.

Information on changes proposed in the new Welfare Reform Bill can be followed on Contact a Family's website. The changes include replacing Disability Living Allowance with the new Personal Independence Payment (PIP), which initially will not be extended to disabled children. A new Universal Credit is being introduced to replace most means-tested benefits and tax credits. Contact a Family is working with other organisations to influence the progress of the Bill as it goes through Parliament. They will also be working with the Department for Work and Pensions to assess the impact on families with disabled children.



Message in a Bottle

The Lions 'Message in a Bottle' encourages people to keep their medical details in a common place (the fridge) where the emergency services expect to find them. This scheme is free to the user and focuses on the more vulnerable people, particularly our children.

As a minimum, it will save the Emergency Services valuable time identifying you and your emergency contacts. By telling whether you have special medication or allergies or not, it is a potential lifesaver and provides peace of mind to users and their friends and families.

Bottles, which are free of charge, can usually be found in your local chemist or doctor's surgery. If you have difficulty in locating a bottle contact the Lions Club at: miab@lions.org.uk.

The scheme is supported by all the emergency services and NHS PCTs.

NHS Safe and Sustainable Children's Congenital Heart Services Programme

It has been recommended that the NHS should develop fewer, larger centres of expertise for children's heart surgery to ensure the best surgical outcomes. At present, 10 centres in England perform heart surgery on babies and children. Spreading surgery across this many centres makes it impossible for all of them to do enough operations to develop skills and expertise to the same level. This has been much in the news lately, with a final decision expected on 14th December 2011.

There have been many debates about which centres should remain open. As a member of the Children's Heart Federation (CHF), BST has focused on putting surgical excellence first as the overwhelming majority of families polled stated that they would be prepared to travel greater distances for better care. CHF is working to ensure that the challenges families face, including problems arising from travelling further, are addressed.

Barth Syndrome Trust Family Gathering

A fun and heart-warming event

By Agnieszka, Wojciech and Sebastian (BTBS Family), United Kingdom



Sebastian and his Dad, Wojciech, enjoy the Family Gathering!
(Photo courtesy of BSTrust ~ 2011)

The Barth Syndrome Trust's Family Gathering on 1st October 2011 was our first and we didn't know what to expect. This great event to which we had been invited was preceded by the Bristol Barth Clinic where we could take advantage of the professional examination of our son complete with echocardiogram, etc., and we could learn more about Barth syndrome from doctors and the medical teams. But the best thing was that we could get to know the families of the other boys with this rare syndrome. We could gain support from other parents and receive hope by seeing and listening to affected families. This is really valuable for a family like ours, especially because we have lived in England for only a few years and all our relatives, family and friends are hundreds of miles away. It is so important to find the strength, support, and understanding. The warmth and friendliness of everyone was overwhelming. The day

at the Avon Valley Country Park was truly a fun and heart-warming event, full of activities for our children and families. We enjoyed great food and one another's company.

I asked my son, Sebastian, what I should write for this newsletter. He looked at me and said, "All you have to write is that it was good fun and really great to meet all those people like me. We all understand each other". We could not ask for more!!! We came home deeply moved. We were met with much kindness and were so pleased with the wealth of information. It was an experience we'll never forget.

To Our Families and Friends...

Thank you, thank you, thank you!

Behind every fundraising effort there is a very special and inspiring person and a story. We would like to thank our families and their friends for their kindness and thoughtfulness in helping everyone affected by Barth syndrome.

Fundraisers

Vincent, age 8, and his twelve year-old sister, Phoebe, assisted by their cousins, Milly (11) and Alfie (8), raised £11 at their seaside 'stall' through donations for their painted stones and shells. Phoebe made posters and wrote a page of information about BST and its importance to her and her family. Vincent told anyone who would listen all about Barth syndrome!

Suzy Green, a new trustee, is a tireless fundraiser, whose latest efforts include a Pampered Chef Party and bucket shaking at a car boot sale. She has raised more than £700 since the last newsletter. Patricia, Suzy's sister, will be running for BST in the London marathon 2012. Friends Therapeutic Community Trust, where Suzy works, once again generously donated the proceeds of their Bonfire Night, £435.

Since we last wrote, we have received an amazing £2100 from Wilf Smith's heroic 80-mile fundraising and awareness walk from Skegness to Sheffield.

Faithful Friends

Thank you to our dependable band of donors whose monthly donations through standing orders so far this year have amounted to £1500 before Gift Aid.

(Cont'd on page 25)

Thank you, thank you, thank you!

(Cont'd from page 24)

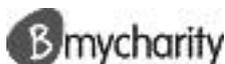
Donations from Europe, UK and Worldwide

'In memory of Oscar and to help others like him'; 'In support of your foundation for Jules and all the other boys'; 'Thanks to NHS Barth Syndrome Service staff, special thanks to Michaela and her family and to Susanna;' 'Brilliant cause!'; '...an excellent charity'. These are some of the very moving comments we have received with donations.

giftaid it

Gift Aid received from the taxman in the tax year ending April 2011 was £1397.00. UK taxpayers, please consider gift aiding your donation and help us reclaim tax you have paid (25p on every pound you donate). You must pay an amount of Income Tax and/or Capital Gains Tax for each tax year that is at least equal to the amount of tax that charities will reclaim on your behalf for that tax year.

Why not donate to BST on line?



www.bmycharity.com provides an easy online donation and fundraising facility. If you are planning a fundraiser you can set up a page, ask people to donate online and track progress. We have a number of sponsorship pages there now. Rachel Farrow arranged a virtual lunch with donations to BST; Kevin and Lizzie Stagg plan to run in the BUPA London 10,000 on 27th May 2012; Eden Amos is swimming 22 miles (equal in distance to the English Channel) over three months and donations have already exceeded his target of £600; Joshua Haycock and team plan a Cycle Ride from Land's End to John O'Groats in August 2012. Hats off to all of you!

Collecting boxes



We are always amazed by our static collecting boxes in UK. They sit on counters in pubs and shops and people drop in their donations — anything from coppers to, on rare occasions, a £20 note. We have had the odd peanut! Even in these difficult times people have been generous and our boxes have together raised over £1,500 so far this year and £8,890 since we began this scheme in mid- 2007. Would you like to raise money and awareness locally and try a box in a friendly shop in your neighbourhood?

Internet shopping?

You can raise money for BST just by doing what you would normally do. Our page at the **Simple Fundraising** website includes many major shops including supermarkets, Amazon and Argos, who will pay a commission to BST for the additional custom. It doesn't cost anything. Go to www.simplefundraising.co.uk/2120 , register your email address and a password, and start shopping.



Swimming Fundraiser: Eden (16) with his brother, Jack (5)
(Photo courtesy of BStrust ~ 2011)



Best of friends, Dillon (5) and distance walker, Wilf Smith
(Photo courtesy of BStrust ~ 2011)

In Loving Memory of Oscar



Oscar
26 October 2010 — 21 June 2011
(Photo courtesy of BStrust ~ 2011)

This is our wonderful son Oscar who some of you may have met back in March of this year in Bristol. As some of you may be aware, Oscar sadly passed away in June this year whilst waiting for a heart transplant in Great Ormond Street Hospital.

In May, a week after we moved house from London to West Byfleet, Oscar became very unwell with breathing difficulties so we rushed him to Kingston Hospital (where Oscar had been born seven weeks early on the 26th October 2010). We were then transferred to Royal Brompton Hospital PICU as he needed more specialist care. We were with Oscar here for about two weeks during which time he began to suffer arrhythmias and we were advised that Oscar's best hope was for a heart transplant. Luckily a bed became available in GOSH so we were transferred with Oscar via the London CATS team (Children's Acute Transport Service).

A couple of days later Oscar had a single chamber Berlin Heart fitted to help him while we began the long wait for a heart. Initially this was a success; but unfortunately Oscar started forming many blood clots and sadly on the 21st of

June one of these travelled to his brain and caused a massive injury that he couldn't recover from.

It is a great comfort in writing this to know that many people who will read it are, or have been in a similar situation to ourselves and understand what we have been through. As Barth syndrome was very new to us when we came down to the Clinic in Bristol we didn't stay very long and have subsequently regretted not staying longer to meet more of you as you were all very friendly and understanding. Some of you pointed out that the Barth boys feel very comfortable in one another's company as they don't have to explain anything to anyone, and we felt the same with all the parents.

We really would like to thank everyone who was involved in Oscar's care, obviously the entire team at Bristol, but also those at Kingston Hospital where Oscar was born, the Royal Brompton Hospital who we were frequent visitors to for checkups and the two week stay before being transferred to Great Ormond Street Hospital, and of course the team there.

Oscar never really looked unwell on the outside, which was a constant surprise not only to friends and family, but also to all the medical people we met. We feel that with the exception of the last three weeks, Oscar had a wonderful life and we are amazed and overwhelmed as to how well he coped.

Finally, thank you very much to Michaela and Annick and everyone at the Barth Syndrome Trust for all their advice in the beginning and their ongoing support. You are a great source of comfort to us.

With best wishes,

Guy, Liz & Oscar in spirit.
United Kingdom

Barth Syndrome Foundation of Canada

President's Report — Winter 2011 Newsletter

By Lynn Elwood, President, Barth Syndrome Foundation of Canada



Lynn and her son Adam (age 21*)
(Photo courtesy of Amanda Clark ~ 2010)

Whenever I sit down to do one of these semi-annual reports, I am reminded just how much work gets done by this organization. We are very lucky to have such a dedicated and efficient group of volunteers working on behalf of all our guys affected by Barth syndrome, and we cannot thank them enough for all their hard work.

In our last newsletter, we mentioned that we were having a Needs Assessment done in order to have an impartial view of the true needs of our member population, especially the men and boys affected by Barth syndrome. We have been very fortunate to have BJ Develle work with our members and their families, and we were so anxious to have the complete report that we asked BJ to join us at our Annual General Meeting and family outreach meeting in April of this year. Thankfully, he made the trip up to the cold north, shared time with the group, and was able to complete the report.

The Annual General meeting itself went well, and we had good attendance from families. In response to a request from our guys, we followed the meeting with an afternoon of bowling. It was a resounding success. Everyone had a great time, and it was wonderful to watch the Barth affected boys and men spending time together and with the siblings. They thanked us for hearing their requests, and despite ending the day very tired, asked that we do something similar for the next outreach.

This was just one of the types of requests we discovered from the Needs Assessment. Several of the findings we believe apply to the larger Barth population, and we will share a summary of these with the International organization. For the areas that are specific to our population, we are acting on delivering as part of our programs. We really have shifted our focus as an organization to the new mission... "Enhancing the lives and outcomes of Canadian individuals and families affected by Barth syndrome," and our members have been vocal in their appreciation.

Other gatherings over the last six months have included our annual golf tournament. This year, we were at a new venue and were fortunate to have four of our guys and some of our Barth friends from the US join us for the day. As you'll see from the article later in this edition, the day was a resounding success in awareness, outreach, and also in raising much needed funds.

The most recent outreach was done in Eastern Canada. Two of our board members and one of our guys travelled to Halifax to meet with doctors, researchers and with one of our Barth young men, Lyem, and his family. This has been one of our strongest awareness events this year, and is a good example of our Barth men taking part in significant ways within the organization.

A few weeks ago, Lindsay Groff joined BSF, Inc. as the new Executive Director. One of Lindsay's first acts was to reach out to the international affiliates, like BSFCa, and learn more about what we do and where we focus. We welcome Lindsay and look forward to working closely with her in our shared global Barth syndrome goals.

Of course, our continued financial health is critical to enable delivery of our programs. As you'll see from the financial summary later in the newsletter, we continue to be in a strong financial position. The conservative budgeting and close watch on expenditures by the board and executive have been key to this success, and, of course, the contributions of all of our Friends of Barth, through donations and participation in fundraisers make it possible. Thank you to all of you for your continued support, in whatever form that takes.

(Cont'd on page 28)

Barth Syndrome Foundation of Canada

President's Report — Winter 2011 Newsletter

(Cont'd from page 27)

Financial Update

While we maintain a continuous and careful eye on our expenses and revenues throughout the year, as year-end approaches we start to look a little more attentively at the figures to see how we have fared financially in the last few months.

We are happy to report that we have been able to follow through money-wise with all of our program plans for the year.

We continue to be very careful in our spending habits. We remain a small but powerful organization, made up entirely of volunteers, and consequently all of the donations and funds that we receive go directly into our programs. With successful fundraisers such as the golf tournament, and direct mail appeals, along with the fantastic profits from fundraisers held on our behalf, we are able to continue to look forward and make plans to carry out our mission and goals, without having to cut back.

Along with the fundraisers that are mentioned in this newsletter, this year we are the happy recipients of a generous contribution from the Max Bell Foundation. This grant-making organization has a mission with emphasis on health and wellness. We are very grateful to Ken Marra, the husband of Susan McCormack, a member of BSF's Board of Directors, for being instrumental in getting the donation for us. Ken and Susan are the parents of a young girl who is a carrier of the Barth gene.

What a pleasure it is to have such great support and to be able to remain positive in a difficult economy. Thanks to all.

Canadian Doctor: Champion of BSFCa

By Cathy Ritter, RN, Vice President and Christine Hope, Treasurer, BSF of Canada



Christopher McMaster, PhD
(Photo courtesy of BSF ~ 2010)

Toward the end of October, with the encouragement and help of Dr. Christopher McMaster, PhD, BSF of Canada participated in a grand rounds meeting at the IWK Children's Health Centre in Halifax, Nova Scotia. Ryan and Cathy Ritter gave a joint presentation to a room full of doctors and other medical professionals.

The enthusiasm of Dr. McMaster's colleagues was encouraging as they were made aware of Barth syndrome, a disorder that they knew little or nothing about.

Ryan covered their family history dealing with Barth syndrome, as well as some of his own problems and successes, followed by Cathy who gave an in-depth explanation of what Barth syndrome is, how it can present in affected individuals, along with current treatments, and most importantly how to diagnose the disorder. Dr. McMaster then gave a brief overview of his ongoing BSF Research Grant project (supported by BSFCa) into finding a drug to help combat Barth syndrome, which was then followed by a question and answer period.

The presentations were very well received, and, we are heartened by the interest that was shown. Thanks go to Ryan and Cathy for taking the time and for being such ideal ambassadors for the Foundation.

We were also able to meet up with Dr. McMaster and a couple of co-researchers in a more informal setting. He was able to tell us that the research grant we funded has led to further major funding from Genome Canada for research in "orphan diseases," including Barth syndrome.

While in Nova Scotia, we took the opportunity to meet up with Lyem, a Barth syndrome affected individual and his mother and grandmother. It was great to be able to re-connect and share recent events and future plans with them and to enjoy some down home eastern friendliness, while catching up with our east coast family.

A Positive Outlook on Life with Barth Syndrome

In Conversation with Robert Hope



Robert enjoys a visit to Royal Tyrell Museum, Drumheller, Alberta
(Photo courtesy of BSFCa ~ 2011)

By Les Morris, BSFCa Volunteer, BSF Publications Team

Attending Barth syndrome functions for seven years has allowed me the great pleasure of meeting these young Canadian men and sharing their progress from childhood to adulthood.

Robert is now twenty-five and employed full time in a position which is like any job — not without challenges.

“I am often required to lift and stack heavy material, and although I am sometimes tired at the end of the day or the end of the week, it’s great to be working. I find that at first although some work is difficult, I gradually become better conditioned and therefore successful.”

Robert says his schooling was relatively normal through elementary, high school and college, sometimes receiving extra help but always with great family support.

“I did have some special interest in history, but after a number of years of working, it is difficult to think about going back to school. If I do go back, I think I would like to be a teacher.”

Personally, I think Robert has already done a lot of teaching. He has made presentations to groups explaining Barth syndrome, has represented our Canadian Foundation at conferences, and he has taken part in many annual general meetings before and after the age of eighteen when he could officially vote on resolutions.

Robert still lives at home, conveniently near to his workplace, with Mom, Dad and brother, Andrew. A favourite hobby of Robert’s is competitive computer games.

In summing things up Robert says, “If and when we find a cure for Barth syndrome, it will be wonderful, but I’m leading a good life. I have a driving license, and I am looking forward to even more independence in the future.” My congratulations to you, Robert. You are a wonderful role model for all the Barth families.

From the Heart — The Two Jans



Jerry & Janet Warren together with Jack & Janet Humphries
golf in support of BSFCa
(Photo courtesy of BSFCa ~ 2011)

By Jan Warren and Jan Humphries

Friends refer to us as the two Jans, and we have been close neighbours in Scarborough for thirty years. When Les Morris was principal at a local school, he also worked with our Block Parent and Community Associations. Friendships developed, and we became acquainted with each other’s families as well. We met Lynn Elwood and her family while visiting at the lake. At that time, they were learning more about their young son Adam’s Barth syndrome symptoms and figuring out and how to help him live with this condition.

With his proud grandparents, we have cheered Adam’s growth since he was a youngster racing over to Grandpa’s for extra bacon (very) early in the mornings. We applauded new

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From the Heart — The Two Jans

(Cont'd from page 29)

achievements like his tractor riding, then driving, woodworking, mastering the outboard and every other glorious machine he came into contact with. Adam's family's dedication to learn more about Barth syndrome, and their ambition to educate others about it, always impressed us.

Since then, we have invited the Barth annual volunteer think tank sessions to take place in our homes. It has provided us with opportunities to meet and hear from other Barth family members and supporters. As ideas are proposed for further promoting educational and medical Barth materials, the brainstorming, networking and enthusiasm have seen many projects come to fruition. The annual golf tournament, silent auctions, coin drives, dances and other fundraisers have become part of our lives. It is exciting to hear the latest achievements and success stories of the medical teams working for Barth syndrome and to learn how the funding is helping the gentlemen and families living with this disorder. That is what keeps us coming back!

Golfers, Good People, Great Course, Gorgeous Day..... Let's Golf!!

By Lois Galbraith, BSFCa Volunteer

Where can a person find great fall weather, good friends, fantastic raffles and unique silent auction items? Add to this four fantastic ambassadors for Barth syndrome (Robert, Adam, Ryan, and Travis), a new and beautiful golf course and scrumptious meals over a few drinks. Yes...you are at the BSFCa annual golf tournament.

Monday, September 12, 2011 did not disappoint any of us. We enjoyed the new venue at Woodington Lake Golf Club (WLGC). We experienced tough but pristine golf holes. We shared the day with dedicated, hard-working volunteers, and we raised \$21,000 for the BSFCa.

Our thanks go out to the record number of golfers who attended our seventh annual golf day. Again this year, our friends Sharon, Jan and Joanie made the trip all the way from Florida, and we helped golfer Lynda celebrate her 50th birthday!!



*Lois, Sharon Olson, and Jan Kugelmann (relaxing)
(Photo courtesy of BSFCa ~ 2011)*



We would like to thank all of the prize donators, the hole sponsors, the numerous volunteers, the loyal golfers, the BSFCa Executive, and especially our Corporate Sponsor, Hope Aero, and our luncheon sponsor, the Buss Megg Society, and of course Jamie, Director of Golf, WLGC, for his generous donation of golf foursomes. Please keep Monday September 10, 2012 free for another great day of golf and comraderie!!

Photo: Our Barth guys: (L-R) Travis, Robert, Adam, Ryan, and Lois (Photo courtesy of BSFCa ~ 2011)

Boogie for Barth — A Repeated Fundraising Success



Lois, George Oliver, and Carol Wilks dance to Mustang Sally (Photo courtesy of BSFCa ~ 2011)

Saturday, October 1st was the chosen date for Susan and Bob McJannett's Boogie for Barth dance, held again at the end of the road in the Toronto Humber Yacht Club on the scenic Humber River. This year, the date was changed from August to October to accommodate the cottagers who had been unable to attend the first one.

There was a wide selection of interesting items in the silent auction (45 in all) that brought some spirited bidding. No matter where your interest lay, there was something to catch your eye. Restaurant certificates, hand made furniture, books signed by the author; unusual chachkees were all up for grabs.

We were treated to George Olliver and his full band "Gangbuster." This year George was supported with a base guitar, drums and keyboards. This allowed him to get out and mix more with the crowd. Again, he brought his "A" game, and the dance floor was jumping.

There was one unusual event during the evening!! One of Bob's friends, Lou Meehan, told him he would pay \$1,000 if Bob would sing. "For \$1,000 for Barth I am willing to make a fool of myself," said Bob. So during the first set, Bob got up and sang the Big Bopper's song Chantilly Lace. "You know it proves that four great musicians can make anyone sound good," said Bob. Lou showed up with his cheque the next day.

Again this year, George had the ladies up singing Mustang Sally. It appeared to us that everyone had a good time. When the dust settled, we raised in excess of \$5,500 for BSFCa, plus we had loads of fun. Hats off to Bob and Susan, Les Morris and Lois Galbraith, Bruce and Carol Wilks plus Frank Malfara and Linda Upshall who showed their dedication to BSFCa by helping with the set-up, rounding up auction items, and just supporting the project. Now what can we do for next year? I hope you will all be there with us.



Bob sings Chantilly Lace (Photo courtesy of BSFCa ~ 2011)

Rick Elwood's 50th Party/Fundraiser

The big day was coming up. Rick was turning 50. We wondered what to do to mark the day. Really the thing Rick wanted most was time with good friends. The idea was born — why not have a party and fundraiser together? So it was that in June, we had one of the best nights of fun with friends and family that we've had in a long time. At the same time, we raised funds for the charity so near to our hearts — Barth Syndrome Foundation of Canada.

We gathered at Grizzly's in Pickering, shared some food and time with friends, and danced the night away to the outstanding "Just Us" classic rock band. It was amazing to see so many people from Rick's work, friends from way back, family, lots of dojo people, and friends from hockey and other sports.

Special thanks to Cheryl, Jeannine and Lois who organized the draws and made sure everyone was invited to participate. Also, special thanks to Steve for making the beautiful bowl for the draw, and to Sensei Jardine who donated his draw winnings and then fittingly won the bowl. Adam, our son with Barth syndrome, joined for part of the evening and acted as the designated driver to get several of us home safely.

Rick's party was very special – the type of warm, friendly and fun night he so richly deserved. It was a terrific night, and raised over \$1,000 for a great cause. Thank you to all of you who joined us.

Welcome Letter from Barth France

By Florence Mannes, President, Association Barth France



Raphaël, age 2
(Photo courtesy of Barth France ~ 2011)

See page 33/35 for the French translation of this article.

Three years ago, we didn't know the meaning of the words "Barth syndrome." Two years ago, we didn't know the Barth Syndrome Foundation nor the Barth Syndrome Trust. One year ago, we only knew a few francophones suffering from Barth syndrome.

What a long way we have come in the last three years, and particularly in the last twelve months.... When we founded Association Barth France in October 2010, we were unaware that only a month later Raphaël would be in a critical state; then we realized fully how important our fight would be.

In November 2011, by communicating about the disease, we learned that 14 cases had been diagnosed in France. Since then, we have been able to contact other families and doctors.

At first, we thought that Barth syndrome was going to isolate us, that we were alone in this fight. When we thought this disease would crush us, it enabled us to search deep down to unknown resources, to become stronger, even better people. Later, first through the BSF and the BST, then in creating Barth France, we met amazing people, devoted men and women, and doctors who were ready to get things moving. The joys of each day now have a particular meaning.

Although we created Barth France initially to raise funds, we now have broadened our projects and objectives:

- Year 1 — Diagnose more families;
- Year 2 — Inform all the teaching hospitals in France about Barth syndrome; and
- Year 3 — Have a French investigator apply to the BSF Research Grant Program,...and so much more!



Members of Ironman4Barth Team
(Photo courtesy of Barth France ~ 2011)

Update on Barth France

Thanks to the involvement in the Nice Ironman of eight triathletes for Ironman4Barth, we were able to raise awareness about Barth syndrome (press articles, television, documentaries...) and to raise funds. Michael finished the 226kms in a little over nine hours while the seven others took between 12 and 13 hours to cross the finish line!

We thought the official tee-shirt of the event "When I grow up, I want to be an Ironman" Raph is wearing in the photo above was quite representative of Barth France: communicating via sports, keeping faith, so that one day Raph and his friends can become real-life Ironmen.

Among the many other sporting events organized by Barth France, we can also mention participation in the five stages of the "Grand prix Lyonnaise des Eaux de Triathlons," where 10,000 leaflets were given out on the disease in Paris, Nice, Tours, Dunkirk and La Baule. In addition, five runners entered the "Course des Héros" in Paris, while five others participated in the Brussels Half Marathon. We also organized a golf tournament for 65 golfers. Finally, triathletes also ran the Aix-en Provence Half Triathlon under the Barth France colors.

There were also less sporty events such as the collection of used clothes, the organization of an information day about Barth syndrome in Raphaël and his brother's school, and also in a school in New York, in which kids drew for children suffering from Barth syndrome. Money was also raised by giving a classical music concert for Barth France.

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Mot de bienvenue de Barth France

By Florence Mannes, Présidente de l'association Barth France



Raphaël, 2 ans

(Crédit Photo de Barth France ~ 2011)

Voir les pages 32/34 pour la traduction anglaise de cet article.

Il y a 3 ans, nous ne connaissions pas le terme même de Syndrome de Barth. Il y a 2 ans, nous ne connaissions pas la Barth Syndrome Foundation, ni la Barth Syndrome Trust. Il y a 1 an, nous ne connaissions qu'une poignée de francophones atteints du Syndrome de Barth.

Que de chemin parcouru au cours de ces trois dernières années, et tout particulièrement au cours des 12 derniers mois... En créant l'association Barth France en octobre 2010, nous ne savions pas qu'un mois plus tard Raphaël serait dans un état très critique, réalisant à quel point notre démarche avait du sens.

En novembre 2011, en communiquant autour de la maladie, nous savons maintenant que 14 cas ont été diagnostiqués en France ; nous avons pu prendre contact avec d'autres familles, avec d'autres médecins.

Alors que nous croyions que le Syndrome de Barth allait nous isoler, que nous étions seuls dans ce combat, nous avons rencontré, d'abord via la BSF et la BST, puis en créant Barth France, des personnalités fantastiques, des hommes et des femmes dévoués, des médecins qui avaient envie de faire bouger les choses. Alors que nous croyions que cette maladie allait nous anéantir, elle nous a permis de puiser au fond de nous des ressources insoupçonnées, de devenir plus forts, parfois meilleurs. Les joies de chaque jour ont maintenant une saveur particulière.

Alors que nous avons créé Barth France initialement pour faciliter la collecte de fonds, nous avons, au bout d'un an, plein de projets et de nombreux objectifs :

- dans 1 an plus de familles diagnostiquées,
- dans 2 ans, tous les CHU de France informés sur le Syndrome de Barth
- dans 3 ans, des chercheurs français candidats à BSF Grant Awards,et bien plus encore!



Les membres de l'équipe Ironman4Barth

(Crédit Photo de Barth France ~ 2011 de Barth France ~ 2011)

Des nouvelles de Barth France

Via leur participation à l'Ironman de Nice, les 8 triathlètes d'Ironman4Barth ont permis de mieux faire connaître le Syndrome de Barth (articles dans la presse, reportage télévisé, ...), et ont permis de collecter des fonds. Michael a parcouru les 226 km en un peu plus de 9h, tandis que les sept autres coureurs ont mis entre 12 et 13 heures pour franchir la ligne d'arrivée!

Nous avons pensé que le tee-shirt officiel de l'épreuve "When I grow up I want to be an Ironman" que porte Raphaël sur la photo représentait assez bien Barth France: Communiquer via le sport, garder l'espoir, pour qu'un jour peut être Raph et ses copains puissent devenir des Ironmen dans la vie,

Parmi les autres événements sportifs organisés par Barth France, il faut également citer la participation aux 5 étapes du grand prix Lyonnaise des eaux de triathlon avec la distribution de près de 10 000 tracts sur la maladie à Paris, Nice, Tours, Dunquerque et La Baule, l'engagement de cinq coureurs à la Course des Héros, à Paris, de cinq autres au semi marathon de Bruxelles, ainsi que l'organisation d'un Trophée de Golf, regroupant 65 golfeurs. Des triathlètes ont également couru le semi Ironman d'Aix en Provence sous les couleurs de Barth France.

Il y a également eu des événements moins sportifs, tels que la collecte d'habits usagés, l'organisation d'une journée d'information sur le Syndrome de Barth dans l'école de Raphaël et de ses frères mais aussi dans une école de New-York, durant laquelle les enfants ont réalisé des dessins pour les enfants atteints du Syndrome ou un concert de musique classique au profit de l'association.

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Update on Barth France

(Cont'd from page 32)

At first, our friends and families helped us create Barth France. As we proceeded, however, there have been so many very enriching encounters with people whom we met at different events, or on the internet.

- There was a mother who lost her son to Barth syndrome five years ago and who wrote us such an encouraging letter.
- There was Xavier, whom we didn't know a year ago, but who has helped Barth France through his communications job.
- There was Virginie, who has planned on running the "Raid Amazone" in Guyana, with two of her friends, to reach out to people about the illness, especially in this French territory far from France.
- There were two French mothers whose sons have Barth syndrome, who, because of the language barrier, could not understand information given out by the BST or the BSF. We have been very happy to communicate with them and translate information given by the BST or the BSF, or learned on the Listserv, and we hope that these families will be able to come to the NHS Bristol Clinic in England.
- There was also Dr. Donadieu, a hematologist, the Head of the French Severe Chronic Neutropenia Registry, whose aim was to list the children in France suffering from chronic neutropenia, and to analyze the use of G-CSF for those children. Through him, we discovered that before December 2009, no case of a child with Barth syndrome had been listed in the register. Since the first case was registered, the work of Dr. Donadieu and his team has enabled the listing of 13 other cases of Barth syndrome (while we only knew about eight cases in France).

We were actually quite surprised to learn that, in very different illnesses, but in which neutropenia is one of the symptoms, some of the same biological anomalies of cell functions are found. This allows researchers to see a new path to investigate, in order to understand better the mechanisms leading to neutropenia. We hope Barth France will be able to work with Dr. Donadieu and his team, to find the specific mechanisms of Barth syndrome via a case study of the sick children.

All along the way, Barth France works alongside a team of doctors (cardiologists, hematologists, endocrinologists, geneticists, nutritionists) whom we rely on to spread information about Barth syndrome, and who, we hope, will work with Barth France and participate in progress through research on Barth syndrome.

For the year to come, we have many projects:

- A large and important team in the Paris Half Marathon and Marathon for Barth France (50 runners for the half, 40 for the marathon);
- Many triathletes listed for the "Triathlon de l'Alpe d'Huez;"
- The creation and sending of an information booklet on Barth syndrome, to all the pediatricians, hematologists and cardiologists (on condition of financial assistance from a company fund);
- A meeting for the French speaking families; and
- Support the work of many French doctors, hematologists, pediatricians, cardiologists, geneticists in screening and research.

The life of Barth France is only starting, still far from the achievements of BSF or BST, but we are trying to move things on as best as we can.

We can't end this article without paying tribute to the doctors of the Necker Enfants Malades Hospital who support us and in Professor Damien Bonnet's words: "Learning by your own experience and that of others — patients, their families and caregivers — are becoming advocates in the management of your illness."

A proud moment for team members of Ironman4Barth
(Photo courtesy of Barth France ~ 2011)



Des nouvelles de Barth France

(Suite de la page 33)

Il y a d'abord eu nos familles et nos amis qui ont participé à la création de cette association. Et puis il y a eu des rencontres, variées et toutes très enrichissantes, des gens rencontrés sur les manifestations ou sur internet

- Il y a eu cette maman, qui a perdu son petit garçon du syndrome de Barth il y a 5 ans et qui nous a écrit une lettre tellement forte d'encouragement;
- Il y a eu Xavier que nous ne connaissions pas il y a un an et qui a mis son métier de la communication au service de l'association;
- Il y a eu Virginie, qui prévoit de courir le Raid Amazone en Guyane, avec 2 de ses amies, pour faire connaître le Syndrome de Barth, en particulier dans ce territoire français loin de la métropole;
- Il a eu ces deux mamans françaises de fils atteints du Syndrome de Barth, qui, du fait de la barrière de la langue, ne pouvaient se rapprocher de BST ou BSF, mais avec qui nous avons été heureux de pouvoir échanger, à qui nous avons pu transmettre certaines des informations communiquées par BST ou BSF, ou apprises sur la L1stserve, et qui, nous l'espérons, pourront venir à Bristol, en Grande Bretagne;
- Et puis il y a également eu ce médecin, cet hématologue en charge du Registre National des Neutropénie, dont le but est de recenser les enfants, sur le territoire français, atteints de Neutropénie Chronique, et d'analyser l'utilisation du G-CSF chez ces enfants;
- Lors de la rencontre que nous avons eu avec ce médecin, le Docteur Donadieu, nous avons découvert qu'avant décembre 2009, aucun cas d'enfant atteint du Syndrome de Barth n'était recensé dans le registre. A partir du moment où un premier cas a été inscrit dans le registre, la démarche proactive du Docteur Donadieu et de son équipe a permis de recenser 13 autres cas de Syndrome de Barth (alors que nous n'avons connaissance que de 8 cas en France).

Nous avons par ailleurs été surpris d'apprendre que, pour dans maladies très différentes, mais dont la neutropénie est un des symptômes, on retrouve certaines similitudes dans les anomalies biologiques au sein du fonctionnement des cellules, ce qui permet d'entrevoir un nouveau champ d'investigation, en vue de mieux comprendre les mécanismes qui conduisent à cette neutropénie. Nous espérons que Barth France pourra collaborer avec ce médecin et son équipe, pour permettre, via une étude spécifique du profil des enfants atteints, de mieux comprendre les mécanismes spécifiques du Syndrome de Barth.

Ainsi, au fil du chemin, Barth France est au contact d'un ensemble de médecins (cardiologues, métabolicien-endocrinologues, hématologues, généticiens, nutritionnistes) sur lesquels il lui est possible de compter pour relayer l'information sur le Syndrome de Barth, et qui, nous l'espérons, pourront collaborer avec l'association, et participer aux progrès de la recherche sur le Syndrome de Barth.

Ainsi, encore beaucoup de projets pour l'année 2012:

- une participation nombreuse au semi marathon, et au marathon de Paris, sous les couleurs de Barth France (50 coureurs pour le semi marathon, 40 pour le marathon);
- plusieurs triathlètes inscrits pour le Triathlon de l'Alpe d'Huez;
- la création et l'envoi d'un livret d'information sur le Syndrome de Barth, à destination de l'ensemble des pédiatres, hématologues et cardiologues Français (sous réserve de l'obtention du financement par une fondation d'entreprise);
- l'organisation d'une rencontre des familles francophones;
- l'implication de médecins français, hématologues, pédiatres, cardiologues, généticiens, dans le dépistage et la recherche.

Nous n'en sommes encore qu'au début de la vie de cette association, encore loin de toutes les réalisations de la BSF ou BST mais nous essayons comme nous le pouvons d'apporter notre pierre à l'édifice.

Nous ne pouvons pas terminer cet article sans rendre un vibrant hommage aux médecins de l'hôpital Necker Enfants Malades qui nous soutiennent dans notre démarche et en reprenant les mots du Professeur Damien Bonnet : " Apprendre par sa propre expérience et s'améliorer avec celle des autres — que l'on parle du patient, de sa famille ou de ses soignants- c'est devenir acteur de sa prise en charge."



Un moment de fierté pour les membres de l'équipe d'Ironman4Barth (Crédit Photo de Barth France ~ 2011)

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Barth Syndrome Foundation

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Christopher (age 3)



Andrew (age 22)

Do you know a boy with this genetic disorder?

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