

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



Henry & Dr. Grzegorz Nalepa are featured in the *Riley Messenger*. See pg. 14. (Photo courtesy of Riley Children's Foundation & Photographer, David Jaynes.)


BSF Research Grant Program An Integral Step Towards Fulfilling our Mission

By Matthew J. Toth, PhD, BSF Science Director

With the completion of the 2011 Barth Syndrome Foundation (BSF) Research Grant Program, ten annual award cycles have committed a total of over US \$2.3 million to this important effort through 64 Research grants to 39 primary investigators. The BSF community should take great pride from the fact that from its beginning, the Research Program of the BSF has been an integral step towards fulfilling its mission. The 2011 Program attracted many fine applications. The BSF, with the advice of its international Scientific Medical & Advisory Board (SMAB), has chosen nine research projects to support financially from this cycle -- the largest monetary commitment by the BSF for any cycle year. The BSF would like to congratulate all the 2011 awardees. See pages 8-10.

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BSF is a proud member of the
National Health Council

Impressions of a BSF Conference Informative, powerful, inspiring and energizing

By Steven M. Claypool, PhD, Assistant Professor, Johns Hopkins University, Baltimore, MD



Dr. Richard Kelley will present at BSF's 2012 International Conference on the "Clinical Aspects & Epidemiology of Barth syndrome."
(Photo courtesy of BSF ~ 2010)

My inaugural Barth Syndrome Foundation conference was in 2008. My life was fairly hectic that summer. My wife had just given birth to our first child. I was closing shop as a post-doctoral fellow and in the process of moving across the country from Los Angeles to Baltimore to start my own laboratory at Johns Hopkins. Sandwiched between the arrival of my daughter and our transcontinental relocation, I attended the 2008 Barth Syndrome Foundation International Conference. When Matt Toth invited me to present my work at the conference I was both excited and apprehensive. For the previous five years in the laboratory of Dr. Carla Koehler at UCLA, I had developed a research project focused on characterizing the yeast version of the protein, TFAZZIN, associated with Barth syndrome. To this point, I had never met another scientist actively working on Barth syndrome. Looking at the list of speakers, I was not only going to meet one other Barth syndrome researcher, but, in fact, I was going to interact with all of the major players in this relatively defined research arena.

(Cont'd on page 4)

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Upcoming Changes to the Barth Registry and Repository

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

After learning more about the Barth Syndrome Foundation (BSF) in the early weeks as Executive Director, it became clear that this organization has celebrated much success. In fact, many of my peers at the National Health Council and elsewhere admire BSF because of how much has been achieved with relatively few resources. Other groups with much larger budgets and a greater number of staff are astonished when they hear about BSF's innovative programs.

The Barth Registry and Repository (BRR) is a particular area where BSF has earned a reputation as cutting-edge. In the first iteration, BSF has paved the way by establishing one of the few rare disease patient registries in existence. Initially, the BRR was operated under an agreement with the University of Florida. Later, it operated in conjunction with Children's Hospital Boston when Dr. Carolyn Spencer, the Principal Investigator (PI) for this project, moved to that institution. To date, this invaluable resource has collected data for 79 confirmed Barth syndrome (BTHS) patients and has gathered biological samples from 45. This is the world's largest deposit of medical information about Barth syndrome, and it will become even more powerful with increased participation.

Many advances have been made in the registry field since the initial creation of the BRR. As such, BSF formed a BRR Committee (consisting of Katherine McCurdy, BSF Board Member, Matthew J. Toth, PhD, BSF Science Director, Shelley Bowen, BSF Director, Family Services & Awareness, and Lindsay Groff, BSF Executive Director) and charged it with evaluating what changes might be needed. The BRR Committee considered numerous options, of which two emerged to be seriously considered for BSF. The first was the current arrangement where the PI is affiliated with the Institutional Review Board (IRB) at his/her institution. The second was to bring the Registry in-house, with a staff member serving as PI and using an independent IRB. The BSF Board of Directors reviewed information about many registries, and on April 18, 2012, with our current PIs' encouragement, voted unanimously to move forward with the second option. We are now making the changes necessary to implement this new and exciting version of our BRR, which we are calling "BRR 2.0."

BRR 2.0 will provide more independence and easier access for researchers because it will be in-house and will utilize an independent IRB (Western IRB or WIRB). Costs should decrease since we will not incur the higher overhead that is associated with academic institutions, and careful planning will minimize interruption caused by changes with the PI. The new platform is focused on patient-entered data, allowing families to be more closely involved in the Registry. Importantly, affected individuals and their families will be more directly able to keep the BRR up-to-date and to influence its success. As the Registry grows, we will be able to quickly and easily obtain summary data about Barth syndrome to help families and researchers learn more.

More information on what families, researchers and physicians can expect will follow in the coming weeks. We look forward to providing detailed information to Conference attendees at the upcoming meeting, including a demonstration about the new platform, and we will make sure that those who cannot attend the Conference will receive the same information. We need the participation of MANY to make this critical resource a real success; this is one of the most direct ways that families can contribute to scientific research and to clinical advances made on BTHS. We wholeheartedly thank those experts and families who got us this far and look forward to this next chapter which we believe will take us far forward.

Thank you for your support as I continue in my Executive Director role; I am honored to be a part of this organization. I see every day as an opportunity to help those who care about BTHS. I can say with confidence that I made the right decision to accept this opportunity, and I hope you feel the same. I cannot wait to meet many of you at the Conference and in your communities!

Barth Registry & Repository

Frequently Asked Questions (FAQs)

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

In order to study any medical condition, it is vital that researchers have data to conduct their studies. With a rare disorder, it is absolutely critical to have a central repository for medical data and biospecimens for that specific condition, since patients are so few and are geographically dispersed.

Given the importance of this subject and the significant details surrounding it, the Barth Registry & Repository Committee developed a Frequently Asked Questions (FAQ) listing below to explain some of the terminology. As we move further into this process, the FAQ will also be expanded and posted to the BSF website to keep all parties fully informed.

Who is on the Barth Registry & Repository Committee (BRR)? Katherine McCurdy, BSF Board Member, Matthew J. Toth, PhD, BSF Science Director, Shelley Bowen, BSF Director, Family Services & Awareness, and Lindsay Groff, BSF Executive Director, comprise the BRR Committee, whose task it has been to evaluate the program and recommend the best way forward. An expert BRR Advisory Board will be established soon to guide the BRR in its creation, operation, and evolution.

What is a patient registry? A patient registry uses observational studies to collect uniform data to evaluate specific questions for those affected by a particular disorder. The Barth Syndrome Registry will allow us to provide researchers an opportunity to study people with Barth syndrome (BTHS), and will help to educate physicians about prognosis and treatment options. Families will benefit from increased research and publication.

What is a repository and why is it important? A repository is a carefully maintained, secure collection of biospecimens (blood, DNA, skin, cell lines, relevant tissue) to be used in research. These samples can be linked with information such as demographic or clinical data about the patient from whom the biospecimen was removed.

Why is participation in the BRR important? The BRR 2.0 offers anonymized data and samples to researchers so that they can more easily increase scientific and medical knowledge about the syndrome. Each individual's medical history and biological samples are important, because 1) each additional data point adds to the aggregate to show a more complete BTHS picture, and 2) each individual is different and may tell an important part of the full BTHS story. We hope that improved knowledge ultimately will lead to clinical advances, which will help all those with the disorder, including BRR participants. Individuals' involvement also will facilitate participation in any relevant clinical trials.

What is new for the participants? Once the new system is up and running, individuals/families will be asked to enter much of their own data on-line (through a special, highly secure portal) so that the medical data are kept up-to-date. Also, adult BTHS individuals or parents of children with BTHS will have the ability to get a report that shows how they/their child compare(s) to others in the registry.

What is a PI? What is his/her role? The Principal Investigator (PI) holds ultimate responsibility for the design, conduct, and management of a research study. Our BSF Science Director, Matt Toth, PhD will fill the role of PI for BSF. Matt will be supported by several others who will help maximize the registry outcomes.

What is an IRB? An Institutional Review Board (IRB) is charged with protecting the rights and welfare of people participating in research.

What is a Western IRB (WIRB)? What are the advantages? It is an alternative to obtaining an IRB through an academic institution. WIRB is a well regarded entity that provides in-depth regulatory expertise to support the development of research protocols and documentation. Choosing WIRB versus having to use an outside PI's own institution's IRB offers timesaving, flexibility and cost saving.

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Impressions of a BSF Conference

Informative, powerful, inspiring and energizing

(Cont'd from page 1)

“...The Barth syndrome research community welcomed me with open arms and was most interested in exchanging ideas and potentially initiating collaborations. We can accomplish much more as a group that communicates than as a collection of isolated research teams. ...”

~ Steven M. Claypool, PhD

Some research fields are extremely competitive and combative. Would the Barth syndrome (BTHS) research community be friendly or adversarial? Would I be worked over like one of my dog's numerous chew-toys? In hindsight, it is amusing to have had such thoughts. The BTHS research community welcomed me with open arms and was most interested in exchanging ideas and potentially initiating collaborations. We can accomplish much more as a group that communicates than as a collection of isolated research teams. Notably, they also forgave me for mispronouncing *tafazzin* and cardiolipin, which I still do. All of the speakers presented a heavy portion of unpublished data. In short, it really seemed as if all of the scientists understood why we perform research on BTHS.

The goal isn't simply to publish high profile papers and get grants (although truth be told, we scientists need quality papers to secure funding and thus have the capacity to continue our research on BTHS). Instead, as a group, we are trying to understand a disease that impacts the lives of real families.

Not surprisingly, prior to the 2008 Barth Syndrome Foundation (BSF) International Conference, I had never been afforded the opportunity to meet the people actually challenged by this disease. Interacting with the BTHS boys and their families and friends both in conversation and in dance was indescribably rewarding. Hearing first-hand about life with Barth syndrome made it abundantly clear that what I do matters and may ultimately make a difference.

My experience at the 2010 BSF International Conference re-enforced my original experience. The scientific sessions were fantastic, my conversations with the other scientists were informative, and every interaction with the boys and families was powerful, inspiring, and energizing.

So what do I hope to gain from participating in the 2012 Conference? I was wondering if I'd be gaining another family member. You see, my first daughter was born just before the 2008 conference, and my second daughter was born just after the 2010 conference. However, unless a stork comes with a really unexpected delivery, it does not appear that my family will be expanding every time I attend a BTHS conference. What I do want from the meeting is to continue to establish collaborations with fellow scientists interested in Barth syndrome and socialize with as many Barth syndrome families as I can. These interactions serve as an important reservoir that helps to sustain us scientists, especially during this incredibly tough funding climate. What we do is important. Just ask the boys and their families.

Editor's Note: Dr. Claypool has been awarded a research grant from the Barth Syndrome Foundation titled “**Characterizing endogenous mammalian TAZ1**” (2011). (See page 8 for a summary of Dr. Claypool's research.) Dr. Claypool will give a presentation at BSF's 2012 Conference on “**The topology of cardiolipin remodeling in yeast**” during the Scientific/Medical Session.

“...I had never been afforded the opportunity to meet the people actually challenged by this disease. Interacting with the Barth syndrome boys and their families and friends both in conversation and in dance was indescribably rewarding. Hearing first-hand about life with Barth syndrome made it abundantly clear that what I do matters and may ultimately

make a difference.” ~ Steven M. Claypool, PhD

Barth Syndrome Foundation Conference

The Highlight of Our Summer

By Brie Chandler-Kalapasev, BTHS Parent, Kentucky, USA



Ned, Brie, Bella & Milosh enjoy hiking!
(Photo courtesy of Brie Chandler-Kalapasev ~ 2012)

In early February 2010 our son, Milosh, was born and went immediately into heart failure. He was only a few days old and in the cardiac ICU fighting for his life, when we first heard of Barth syndrome. We were told that once he was stable and out of immediate danger, he would be tested for it. Nearly two months later, after many ups and downs, it was confirmed — Milosh has Barth syndrome.

We suddenly had answers to our questions. Well, most of the questions. The big “why?” was answered, but many more questions appeared and still do, sometimes on a daily basis. The Barth Syndrome Foundation (BSF) instantly became our lifeline. The BSF website and the Listserv communications and archives were our daily tools that kept us going through some very tough times. When we found out that the BSF conference was happening in summer of 2010, we hoped Milosh would be healthy enough to attend. As Milosh improved, we were cleared to fly and attend the BSF conference.

The 2010 BSF conference was amazing! Both the family and scientific sessions were not only very helpful and insightful, they were vital to our understanding of what this condition is and how it affects individuals and their families. To this day, we still refer back to conference presentations in trying to better understand this very complex disorder. We learned a lot about Barth syndrome itself. We met the medical experts working on finding a cure, participated in medical sessions and assessments, and instantly made life-long friends.

Meeting families and individuals affected by Barth syndrome for the first time was profound, emotional, happy, and comforting at the same time. Until you have experienced it for yourself, it would be difficult to describe how it feels to meet strangers from around the world who understand and know you; they know your deepest worries and fears, and understand your greatest joys in an instant. It is amazing how quickly one becomes part of our BSF family. The camaraderie and support is simply unwavering. Becoming a member of this group is not what any parent wants and hopes for their son and their family. Earning honorary degrees in cardiology, hematology, biochemistry, neurology, or becoming a constant advocate and a teaching tool is not anyone’s idea of parenthood either, but if this is your path, I cannot think of a better, more passionate group of people to join you on your journey.

The most compelling reason, by far, to attend a Barth conference is being able to witness, first-hand, how amazing these boys and men are and how strong they are as individuals and as a group. To say that they share a bond would be an understatement. These guys are beyond any common bond definition. They all have amazing insight and wisdom, lots of appreciation and kindness, and are so much fun to be around. This unique “gift” gained by attending the conference will be the highlight of our summer.

When our family was asked to write an article about what it was like for a recently diagnosed family to attend a Barth conference for the first time, it was suggested that we include the citation to the peer-reviewed medical journal article recently published about Milosh. It was challenging for us to figure out where, in this story, it belonged so we saved it for here (**Left Ventricular Noncompaction Cardiomyopathy in Barth Syndrome: An Example of an Undulating Cardiac Phenotype Necessitating Mechanical Circulatory Support as a Bridge to Transplantation.** *Pediatr Cardiol.* 2012 Mar 17.) Please see page 11 for the full citation and link to the abstract.

And, we would like to take a final moment to say that while the 2010 Conference Friday Night Social was spectacular, the 2012 Conference Friday Night Social will be **LEGEND**..wait for it...**ARY!!!!** Can’t wait to see you all in Florida!

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).

In addition to making your hotel reservation, you will need to register for this Conference with the Barth Syndrome Foundation. Registration is 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



(Photo courtesy of Dr. Groft ~ 2011)

Keynote Speaker

Stephen C. Groft, PharmD, Director, Office of Rare Disease Research, National Institutes of Health, Bethesda, MD, USA

"A Globalization of Rare Diseases Research Activities"

BSF is very pleased to announce that the Director of the Office of Rare Disease Research (ORDR) at the National Institutes of Health (NIH), Stephen C. Groft, PharmD, 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 group has been very generous in financially supporting our biennial conferences. As of December 23, 2011, the ORDR is now a part of the National Center for Advancing Translational Sciences at the NIH. 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.

The Barth Syndrome Foundation is pleased to announce that the Scientific and Medical Sessions of the 2012 Conference is funded in part by grants from the Office of Rare Diseases Research and the National Heart, Lung and Blood Institute of the National Institutes of Health.



Questions and Answers from the Community



By David Axelrod, MD., Pediatric Cardiologist, Lucile Packard Children's Hospital at Stanford, in Palo Alto, California; Board of Directors, Barth Syndrome Foundation

Q What is left ventricular non-compaction (LVNC), how does LVNC relate to Barth syndrome, and what are the three most important health concerns with this condition?

A Left ventricular non-compaction (LVNC) is a rare genetic cardiomyopathy. Most experts define LVNC as a thickened myocardium consisting of two separate muscle layers which create a “spongy” appearance on echocardiogram. The non-compacted layer features deep recesses within the trabeculations of the left ventricular muscle. (Normally, the left ventricle has fine comb-like muscle bands called trabeculations; LVNC describes abnormally thick trabecular bands.)

Barth syndrome is recognized as a genetic disorder which can feature cardiomyopathy; approximately 50% of Barth syndrome patients may meet diagnostic criteria for LVNC. The clinical signs and symptoms of LVNC vary; some patients are asymptomatic while others develop complications from heart failure, abnormal heart rhythms (arrhythmias), or clots (thromboemboli). Treatment focuses on managing cardiac dysfunction and heart failure symptoms, diagnosing and treating arrhythmias, and preventing clot development.



By Audrey Anna Bolyard, Clinical Manager, Severe Chronic Neutropenia International Registry; David C. Dale, MD, Professor of Medicine, University of Washington, Seattle, WA

Q How do I know if my son has neutropenia? How do I know if he has episodes of neutropenia?



A Neutropenia is a condition of lower than normal neutrophils (absolute neutrophil count, ANC) in the blood. Neutrophils are very important in defending the body against bacterial and fungal infections. The normal neutrophil count is between 1.800 to 7.000 x 10⁹/L. Neutropenia is described as mild, moderate or severe; mild neutropenia is when the ANC is between 1.000 to 1.500 x 10⁹/L, moderate neutropenia is when the ANC is between 0.500 to 1.000 x 10⁹/L and severe neutropenia is when the ANC is below 0.500 x 10⁹/L.

Clinical signs of neutropenia are varied. For some people it will be profound fatigue or swelling/bleeding of the gums or mouth ulcers/sores. Other clinical signs of neutropenia are repeat infections that require oral antibiotics to heal such as a repeat skin infections around a J or G tube or it could be a serious infection requiring hospitalizations and intravenous antibiotics.

The best method to discover if your son has neutropenia and/or the severity of neutropenia is to do the serial complete blood counts (CBCs, also called full blood counts -- FBC) with a differential. The physician will use the CBC evaluation to determine the ANC and, along with the infection history, determine if the neutropenia is mild, moderate or severe.

Serial blood counts done each year will help the physician know if the patient is developing neutropenia. Four CBCs (two in one week, such as Monday and Thursday and two in the next week) will give the physician the information needed to determine if the patient is experiencing neutropenia. If in those two weeks the patient is found to be neutropenic the physician may choose to continue the CBCs for 6 to 8 weeks to document the lowest ANC.

(Photos courtesy of Dr. David Axelrod, Audrey Anna Bolyard and Dr. David Dale ~ 2012)

Science Corner

BSF 2011 Research Grant Cycle

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



Steven M. Claypool, PhD, Assistant Professor
Johns Hopkins University, Baltimore, MD
“**Characterizing endogenous mammalian TAZ1**”
Award—US \$40,000 over 1-year period

Characterization of an essential tool for research: monoclonal antibodies against human tafazzin. Dr. Claypool has developed three monoclonal antibodies against the human *tafazzin* protein and shown evidence that at least one of them can detect the protein directly in simple cell extracts. These antibodies will be used to analyze the expression of this protein in different cell types and under different physiological conditions—an ability that the research community has not had before. The research community has been hampered in its ability to easily visualize the *tafazzin* protein which has prevented progress. Dr. Claypool has developed these monoclonal antibodies which show evidence that they will satisfy the requirements of the entire research community. Because monoclonal antibodies are produced using cell culture techniques (hybridomas), there can be an almost limitless production of this vital tool far into the foreseeable future of Barth syndrome research. Specifically, Dr. Claypool will: (1) map which parts of the *tafazzin* protein interact with the antibodies (epitope mapping), (2) identify the subcellular localization of mammalian *tafazzin* protein in human fibroblasts and mouse tissues using confocal microscopy, (3) determine the submitochondrial localization using mitochondrial fractionation and protease-protection assays, and (4) visualize the macromolecular assemblies with other protein supercomplexes using blue-native electrophoresis.



William T. Pu, MD, Associate Professor
Children's Hospital Boston, Boston, MA
“**Using pluripotent stem cells and modified RNAs to model and correct Barth syndrome**”
Award—US \$40,000 over 1-year period

Differentiating iPS cells into cardiomyocytes and correcting the biochemical phenotype of tafazzin gene dysfunction with modified RNA. Dr. Pu will build upon his construction of induced pluripotent stem (iPS) cell lines from two Barth syndrome (BTHS) individuals—a notable accomplishment stimulated by his 2009 BSF Research Grant. The most developed cell line will be differentiated into beating cardiomyocytes to study in detail using various biochemical real-time analytical procedures including mitochondrial function-morphology and pertinent signal transduction pathways. Dr. Pu has provided significant unpublished data, and in collaboration with Dr. Kenneth R. Chien (Director of the Massachusetts General Hospital Cardiovascular Research Center), he will prepare modified RNAs to reverse the BTHS biochemical phenotype in cellular models and in the *tafazzin* knockdown mouse line. This modified RNA technique does not use viral systems but rather small lipid droplets called liposomes to deliver the RNA to the target cell or tissue. The RNA-liposome technology is designed to produce quick but temporary answers to questions about *tafazzin* deficiencies, and for the knockdown mouse experiments it will focus on remediating heart tissue abnormalities.



Mindong Ren, PhD, Assistant Professor
New York University School of Medicine, New York, NY
“**Drug repositioning for Barth syndrome**”
Award—US \$40,000 over 1-year period

Bezafibrate, resveratrol, and N-acetylcysteine for the reversal of cardiolipin abnormalities in tafazzin dysfunctional cells. Dr. Ren has discovered that two compounds when incubated with either *tafazzin* deleted mouse cells or with fibroblasts from Barth syndrome (BTHS) individuals, are able to reverse the monolysocardiolipin/cardiolipin ratio that is distinctive of this genetic disorder. Both compounds also increased oxygen consumption which is a characteristic of many mitochondrial disorders. Because bezafibrate is already a pharmaceutical used in Europe for treating high triglyceride levels and has a 20+ year history of safe use, it is reasonable to project that bezafibrate could be useful in treating BTHS individuals. Resveratrol is currently in clinical trials and is touted as an important ingredient of red wine that provides health benefits. N-acetylcysteine is FDA approved as a mucolytic to treat excess lung secretion, and is known to reduce reactive oxygen species (ROS) which is reminiscent of some of the claims for resveratrol. Dr. Ren intends to treat the knockdown mouse model with these three compounds and monitor for changes in: (1) protein oxidation, (2) phospholipids including cardiolipin, (3) mRNA expression using microarrays, (4) *tafazzin* protein levels, (5) histological analysis, and (5) various physiological parameters including echocardiography and stress echocardiography. If the results are positive, then experiments using cell lines from various BTHS individuals will be screened for similar effects.

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(Cont'd from page 8)



Miriam Greenberg, PhD, Professor and Associate Dean

Wayne State University, Detroit, MI

“Cardiolipin deficiency leads to defects in the TCA cycle”

Award—US \$40,000 over 1-year period

**Funding for this award was provided by the Barth Syndrome France*

Identifying tricarboxylic acid cycle defects in yeast cardiolipin mutants. Building on the tricarboxylic acid (TCA) cycle dysfunction hypothesis of Barth syndrome (BTHS) as put forth by Dr. Richard Kelley, Dr. Greenberg will examine the TCA cycle in yeast mutants that are compromised in their cardiolipin expression, which includes the *tafazzin* deletion strain. Dr. Greenberg has provided unpublished data to show that mitochondrial dysfunction caused by cardiolipin alterations involves and impacts the proper functioning of the TCA cycle—the pivotal metabolic system of mitochondria-containing cells. Dr. Greenberg will measure: (1) metabolite levels, (2) enzyme activities, (3) mitochondrial retrograde pathway gene expression (the pathway of proteins and metabolites whereby the mitochondria communicate with the nucleus of the cell to alter metabolism (*i.e.*, *tafazzin*), (4) TCA cycle enzymes, (5) TCA cycle intermediates, etc.), (6) beta-oxidation pathway (metabolism of fats), and (7) the glyoxylate cycle (a short-circuit of the TCA cycle not found in animals but found in yeast). In addition, the supplementation of oleic acid to the growth media rescues yeast cardiolipin mutant strains, and uncovering the basis for this rescue will be investigated. Dr. Greenberg hopes that by identifying TCA cycle abnormalities we will better understand how anapleurtoic/nutritional supplements (like arginine) may be beneficial in the treatment of BTHS.



W. Todd Cade, PT, PhD, Assistant Professor

Washington University School of Medicine, St. Louis, MO

“Effects of resistance training on cardiac, metabolic, and muscle function and quality of life in Barth syndrome”

Award—US \$39,937 over 1-year period

Resistance exercise training in Barth syndrome individuals. The limited (n = 3) clinical data that was supported by Dr. Cade’s 2009 BSF Research Grant and partially reported here in this application has shown marginal if any improvement in the peak oxygen maximum or in the work rate after three Barth syndrome (BTHS) individuals completed a 12-week cardiovascular training program that focused on endurance training (cardiovascular rehabilitation). Though quality of life surveys and anecdotal reports demonstrated value, it was disappointing not to see a larger physiological improvement after these exercise sessions, however the number of data sets collected is very small and any conclusions are preliminary. In response to this development, Dr. Cade has proposed to shift the type of exercise training and preferentially stimulate glucose-utilizing muscle fibers (non-oxidative, type II fibers, or white muscle) which is compatible with what we know about the energy predilection of BTHS—*i.e.*, the tissues of BTHS individuals do not extract oxygen efficiently which may explain/limit their ability to utilize fat (oxidative metabolism) as an energy source. Dr. Cade has proposed a similar program to what was performed for his 2009 BSF Research Grant. Three BTHS individuals will undergo supervised resistance exercise training (pump you up!) and will be monitored for effects on safety, echocardiographic changes, skeletal muscle strength and mass, whole-body protein synthesis rate, and quality of life. An additional aim will be to monitor arginine synthesis and breakdown rates in these individuals, as it relates to the TCA cycle hypothesis of Dr. Richard Kelley.



Grant M. Hatch, PhD, Professor

University of Manitoba, Winnipeg, Manitoba, Canada

“MLCL AT-1 elevates cardiolipin and mitochondrial function in cardiac myocytes of *taz* knockdown mice”

Award—US \$40,000 over 1-year period

**Funding for this award was provided by the Barth Syndrome Foundation of Canada*

Using lentiviral expression of MLCL AT-1 to correct the *tafazzin* knockdown mouse. Using his discovery that monolysocardiolipin acyltransferase 1 (MLCL AT-1) alters the cardiolipin profile of cells, Dr. Hatch will use lentiviruses to deliver this gene into isolated cardiomyocytes from the knockdown mouse model of Barth syndrome (BTHS). Dr. Hatch’s hypothesis is that genes other than *tafazzin*, like MLCL AT-1, impact the cell’s cardiolipin profile, and he has published that overexpression of human MLCL AT-1 in lymphoblasts from BTHS individuals elevated cardiolipin levels and complex II activity (one of the energy yielding protein complexes found in the mitochondrial electron transport chain). Dr. Hatch will isolate ventricular myocytes from *tafazzin* knockdown mouse hearts and measure gene expression as well as the enzymatic activities of proteins involved with lipid metabolism. Lentiviruses containing MLCL AT-1 minigenes will be prepared in collaboration with Dr. Sam Kung (Associate Professor at the University of Manitoba) and then used to transduce individual cardiomyocytes obtained from the knockdown mouse. These transduced cells will then be analyzed for cardiolipin changes and for gene expression differences. In addition, the mitochondrial function of the transduced cells will be monitored with a

(Cont'd on page 10)

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state-of-the-art machine (Seahorse Extracellular Flux Analyzer) to define the impact on mitochondrial metabolism. Another longer term goal is to transduce the knockdown mouse itself using the same lentivirus technology and to monitor for any physiological or biochemical changes.



Yana Sandlers, PhD, Assistant Director of Biochemical Genetics

Kennedy Krieger Institute, Baltimore, MD

“Characterization of biochemical abnormalities in Barth syndrome patients and mouse model of BTHS”

Award—US \$21,065 over 1-year period

Tricarboxylic acid cycle intermediates in the *tafazzin* knockdown mouse and in clinical samples. In order to support the TCA cycle hypothesis as a mechanism of Barth syndrome (BTHS) pathology, Dr. Sandlers will measure compounds that occur in the blood and urine of BTHS individuals and also in the knockdown mouse model treated with high or low protein diets. Dr. Sandlers presented unpublished clinical data to support her plan to take advantage of the 2012 BSF International Conference and obtain: (1) 3-day prior food records, (2) 5-hour fasting plasma amino acid and TCA cycle intermediate levels, (3) clinical questionnaires, and (4) medical records from BTHS volunteers. Similar but more extensive experiments with the knockdown mouse model will be used to determine if the same perturbations found in humans are reflected in the knockdown mouse model of this human disease.



Ji Zhang, PhD, Assistant Project Scientist

University of California at San Diego, San Diego, CA

“Functional characterization of a mitochondrial lipid phosphatase that involves cardiolipin biosynthesis”

Award—US \$40,000 over 2-year period

Using PTMP1 muscle knockout mice with cardiolipin abnormalities as an animal model to investigate the common pathology with Barth syndrome. Dr. Zhang will use her muscle knockout mouse line of the gene for phosphatidylglycerol-phosphate phosphatase (PTMP1—the complete knockout is embryonic lethal) to identify what pathologies are present in addition to the cardiolipin abnormalities already observed. The PTMP1 enzyme is part of the cardiolipin biosynthetic pathway, is localized to the inner membrane of the mitochondria like the *tafazzin* protein, and belongs to a large family of related proteins called phosphatases (PTP superfamily). Specifically, Dr. Zhang will carry out the following biochemical and physiological studies on her mouse line or on isolated cardiomyocytes from her mouse line: (1) phospholipid composition, (2) mitochondrial morphology, (3) apoptosis, (4) autophagy, and (5) cardiac-skeletal muscle functions. Preliminary data show that the PTMP1 muscle knockout mice have a lower body weight, a higher mortality rate, and an increased heart weight suggesting cardiac hypertrophy. In collaboration with Dr. Anne Murphy (Associate Adjunct Professor at University of San Diego), she will investigate the mitochondrial function using whole cell analysis (Seahorse Extracellular Flux Analyzer). Investigating another mouse model with a cardiolipin abnormality has advantages in identifying what common mechanisms lead to similar pathologies.



Junhwan Kim, PhD, Research Associate

Case Western Research University, Cleveland, OH

“Causative and correlative role of cardiolipin on integrated mitochondrial function in Barth syndrome”

Award—US \$40,000 over 1-year period

Oxidative phosphorylation and electron transport chain measurements in mitochondria from *tafazzin* knockdown mice. Dr. Kim will perform measurements on the mitochondrial function of tissues isolated from the mouse model of Barth syndrome to uncover what parts of the oxidative phosphorylation pathway are affected by this gene dysfunction. Dr. Kim proposes to perform an in-depth analysis of the bioenergetic function of cardiac mitochondria using techniques that are standard in the highly-regarded mitochondrial disease research laboratory (Charles Hoppel Laboratory) of which he is a member. Specifically, Dr. Kim will examine complexes III and IV of the electron transport chain to determine if abnormalities in these multi-protein complexes can be responsible for the increased reactive oxygen species (ROS) often found in mitochondrial diseases.

The funding of these grants will foster a better understanding of Barth syndrome (BTHS) in both a scientific and a clinical manner by providing funding for basic science and clinical research on the natural history, biochemical basis, and treatment of BTHS. Since the 2002 season, the Barth Syndrome Foundation (BSF), in consultation with its Scientific and Medical Advisory Board and with the support of international affiliates (Barth Syndrome Foundation of Canada, Barth Syndrome Trust, UK and Europe, and Barth France), has awarded over US \$2.3 million in research grants in order to better understand this rare genetic disease characterized by cardiomyopathy, growth delay, muscle hypoplasia, neutropenia and extreme fatigue. This competitive grant program has resulted in many publications which are unraveling the details of this multi-faceted disease and are leading towards new ideas for treatment. Many of the past and present grant awardees will be attending BSF's Scientific, Medical and Family Conference on June 25-30, 2012 in St. Pete Beach, Florida.

Awareness of Barth Syndrome Continues to Grow

There has been a significant increase in Barth syndrome (BTHS) related peer-reviewed journal articles published. To date, there has been a total of **63** 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.

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5. Schild L, Lendeckel U, Gardemann A, Wiswedel I, Schmidt CA, Wolke C, Walther R, Grabarczyk P, Busemann C. [Composition of molecular cardiolipin species correlates with proliferation of lymphocytes](#). Experimental Biology and Medicine EBM.2011.011311; published ahead of print 4 April 2012, doi:10.1258/ebm.2011.011311 (Abstract)
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7. Hanke SP, Gardner AB, Lombardi JP, Manning PB, Nelson DP, Towbin JA, Jefferies JL, Lorts A. [Left ventricular noncompaction cardiomyopathy in Barth syndrome: An example of an undulating cardiac phenotype necessitating mechanical circulatory support as a bridge to transplantation](#). Pediatr Cardiol. 2012 Mar 17. [Epub ahead of print] (PubMed Abstract)
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9. Joshi AS, Thompson MN, Fei N, Hutteman M, Greenberg ML. [Cardiolipin and mitochondrial phosphatidylethanolamine have overlapping functions in mitochondrial fusion in Saccharomyces cerevisiae](#). J Biol Chem. 2012 Mar 20. [Epub ahead of print] (PubMed - Free Article)†
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11. Greenberg, ML. [Functions of cardiolipin as modifiers of the Barth syndrome phenotype](#). ASBMB Today. March 2012. (ASBMB Abstract)
12. Cosson L, Toutain A, Simard G, Kulik W, G Matyas, Guichet A, Blasco H, Maakaroun-Vermeesse Z, Vaillant MC, Le Caignec C, Chantepie A, Labarthe F. [Barth syndrome in a female patient](#). Mol Genet Metab. 2012 May;106(1):115-20. Epub 2012 Jan 24. (PubMed Abstract)
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14. Rubinstein YR, Graft SC, Chandros SH, Kaneshiro J, Karp B, Lockhart NC, Marshall PA, Moxley RT 3rd, Pollen GB, Miller VR, Schwartz J. [Informed consent process for patient participation in rare disease registries linked to biorepositories](#). Contemp Clin Trials. 2012 Jan;33(1):5-11. Epub 2011 Oct 24. (No PubMed Abstract available)
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Barth Syndrome Researcher Receives National Institutes of Health R01 to Further Studies on Barth Syndrome

The Barth Syndrome Foundation (BSF) is pleased to share with our community that W. Todd Cade, PT, PhD, has received notice of an award from the National Heart, Lung and Blood Institute of the National Institutes of Health for his R01 grant entitled, “**Heart and Skeletal Muscle Metabolism, Energetics and Function in Barth Syndrome.**” This award is distributed over five years and is in the amount of US \$1.6 M. This is a significant achievement for which the BSF is very proud and excited. Dr. Cade will begin recruitment for this research project at BSF's 2012 Conference!

Funding Opportunities Relevant to Barth Syndrome Research

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

National Institutes of Health (NIH)

Health Promotion for Children With Physical Disabilities Through Physical Activity and Diet: Developing An Evidence Base (R01)

Funding Opportunity Announcement (FOA) Number: PAR-11-288

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

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

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

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

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

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

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

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National Institutes of Health (NIH)

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.

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)

Raising Awareness ~ An International Initiative

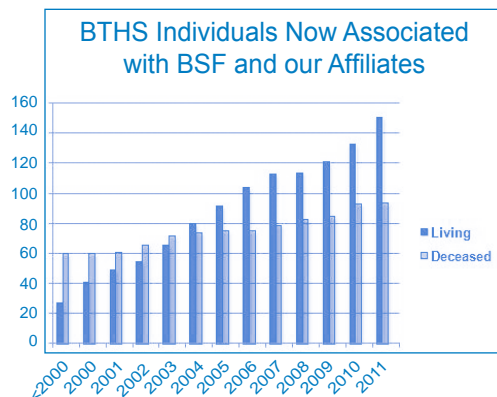
By Shelley Bowen, Director, BSF Family Services & Awareness; Tiffini Allen, BSF Volunteer; Michaela Damin, Chair, Barth Syndrome Trust; Lynn Elwood, President, Barth Syndrome Foundation of Canada; Florence Mannes, Chair, Association Barth France

INTRODUCTION

By Shelley Bowen, Director, BSF Family Services & Awareness

It has long been our belief that early diagnosis is key to survival for those who have Barth syndrome (BTHS). Over the past 12 years we have learned that 90% of all mortality of those with BTHS occurs in the first decade of life. The greatest ratio of mortality within that decade occurs in the first year of life, closely followed by loss during pregnancy. Education and support about BTHS remains an integral priority to save those who have BTHS. Our first priority lies in advocating for an accurate diagnosis. Barth syndrome is never too rare to consider as a diagnosis. Through our awareness efforts we are finding and helping affected families around the world. Your belief in our mission provides us with the resources to do just that.

We are united in one central mission. Our efforts have not been splintered because the mission is as relevant today as it has been over the past decade.



Note:

BSF was founded in 2000 and has contributed to these improved statistics.



Devin and Christopher thank those who have donated in their honor and in support of BSF's 2012 Conference.
("Henry Cards" courtesy of Tiffini Allen ~ 2012)

FAMILIES RAISING AWARENESS IN THE US

By Tiffini Allen, BTHS Parent, BSF Volunteer

"Henry Cards" have made their way across the States and around the globe. The campaign was introduced this spring to raise funds for the 2012 Barth Syndrome International Scientific, Medical and Family Conference. This new initiative is similar to a letter campaign, but rather than receiving a donation letter in your mailbox, a unique Henry Card with a wonderful photo of a boy(s) with Barth syndrome (BTHS) on the front of the card will replace the letter. Each card has a personality all its own. A handful of people have ventured out to see what kind of response they would receive from their family and friends. The campaign has proven to be a huge success as donations continue to pour in. Henry Card e-mail campaigns have also shown to be successful and are just as easy to implement.

It's simple. Send me a photo that you would like to use on the front of the card and I will e-mail you your personalized card within a couple of days. If you don't have access to a printer, I will print the cards and send them to you. You then mail out the cards to your family and friends and anxiously await the arrival of your donations! We would encourage everyone to participate in the Henry Card campaign at least once. We can guarantee that the recipient of your Henry Card will cherish it. If you or someone you know would like to send out your own Henry Cards or have questions, please contact me at tiffiniallen@gmail.com. Thank you to everyone who has participated in our first annual Henry Card campaign!

A SPECIAL RELATIONSHIP

We knew there was something special between Henry and Dr. Grzegorz Nalepa (referred to as Dr. Grzegorz), Henry's hematologist, when they first met in 2008. Although Henry's respiratory rate unfortunately would go up when anyone walked into his hospital room,

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Henry and Dr. Grzegorz during their interview on NEWS 13 WTHR. (Photo courtesy of Tiffini Allen ~ 2012)

it didn't go up the many times that Dr. Grzegorz visited him. Their unique relationship has grown over the past couple of years. Henry loves going to Riley Hospital for Children to visit Dr. Grzegorz even if it involves getting his blood drawn. We needed to thank Dr. Grzegorz for everything that he had done and was doing for Henry and our family, and a thank you letter just wasn't enough. We decided to create a hardcover book explaining their relationship and expressing what a kind and wonderful doctor Dr. Grzegorz is. The book is filled with 20 pages of photos of the two of them. When the Riley Children's Foundation heard about the special bond between Dr. Grzegorz and Henry, they asked if they could share our story in the spring *Riley Messenger*, the hospital's magazine. (Read the entire article at http://www.rileykids.org/stories/archives/henry_allen-dollard/.)

The article in the *Riley Messenger* came out a few days before National Doctor's Day. As a result of the article and National Doctor's Day, Henry and Dr. Grzegorz shared their story and created awareness for BTHS while being interviewed on the radio by Terri Stacy of WIBC, as well as a

television interview on WTHR's Health Beat with Scott Swan. We would like to thank Dr. Grzegorz, Riley Children's Foundation, Terri Stacy, and Scott Swan for sharing our story and educating the public about BTHS.

RAISING AWARENESS IN CANADA

By Lynn Elwood, President, Barth Syndrome Foundation of Canada

In Canada, the BSFCa raises awareness in a number of ways. First, we use every fundraising event as an awareness opportunity. Each year, we have a Golf Tournament and always get new people joining that event. Part of what we provide to the golfers is a small awareness item, something they will wear or use in public and others will see. Examples have included hats, backpacks, patches, car decals, reusable cloth bags and coolers among other things. Also at events like this, we include a copy of our Canadian Newsletter. This publication focuses on the activities of the local organization and always spotlights families, affected boys and men, and volunteers. It is very well received and helps to get people connected to the organization. A number of people have become involved with us after joining our fundraisers or reading our newsletter.

Over the years, we have done a number of mailings to hospitals and doctors to raise awareness. From these, we receive a few contacts and in some cases have had doctors reach out to say they were treating someone with BTHS. We still do this periodically, but we find it more effective to have in-person meetings with physicians whenever possible. Recently, we were able to travel to Eastern Canada where one of the Barth syndrome researchers, Dr. McMaster, organized a group to meet with us. On this trip, we were able to share information about the condition with over 50 physicians and scientists in one meeting. There were some more in-depth meetings with physicians as well. These opportunities are rare, as they require an advocate in the medical or scientific community to promote the event and assist with local logistics, but they are invaluable. We are hoping to do the same thing in the west of Canada.

We do sometimes attend scientific/medical conferences and have a booth, but we choose these carefully due to cost. We run regular ads in the *Medical Post*, which we have secured at no charge, thanks to the generosity of a family member of one of our volunteers. We have volunteers that make Barth Bears that people bring with them when they travel (watch for them at the conference in June). We give educational pamphlets to families so they can carry them and share with interested parties. One of the most important aspects of awareness — we talk about Barth syndrome. All our volunteers and our families share the word as they talk with people, and it is amazing the types of things that result from these conversations. You just never know what can come of a simple conversation. It all helps.



Joe (Photo courtesy of BSTrust ~ 2012)

FAMILIES RAISING AWARENESS IN THE UK

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

In April, one of our boys, young Joe, made international news. After the story broke on ITV national news in the UK, it quickly spread with coverage in all the main newspapers and television stations.

Joe spent a record 251 days attached to an artificial heart while he waited for a transplant, and it was in November last year that he finally received his donor heart. His parents, Rachel and Mark made a heartfelt appeal for more people to sign up to the donor register saying, "We are eternally grateful to the donor family. We cannot imagine what they went through. Their generosity of thought at such a horrendous time is completely selfless and amazing."

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Raising Awareness ~ An International Initiative

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The footage of little Joe saying, "I've got a new heart now, I've got a little one," brought a smile and quite a few tears to all who watched, especially for those within our community who have been supporting Joe and his family throughout this long and uncertain time. We wish you all the best little man! (For the full story, go to <http://www.itv.com/news/2012-04-10/family-appeals-for-donors-after-a-new-heart-saved-their-sons-life/>.)



BSTrust Awareness Booth at the Hampshire Special Needs Info Day
(Photo courtesy of BSTrust ~ 2012)

Special Needs Information Day at the Petersfield School, Hampshire

Helen Coleman, trustee and mother, spent a day raising awareness at this well-attended event organised by Contact a Family and Hampshire Parent Partnership Service. This annual event gives families and practitioners the opportunity to gather information about support and services available across the county through a series of workshops and seminars. Many people came to visit the BST stand and chat to Helen throughout the day.

UK Rare Disease Plan

In February, the UK launched a collective public consultation on a UK plan for rare diseases on Rare Disease Day. In May, Ralph Easterbrook, volunteer and father, joined Dr. Vanessa Garratt and Debbie Riddiford from the Bristol Service to take part in this meeting.

The importance of good communication and dissemination of information emerged as vital to ensure speedy diagnosis and highly co-ordinated care between specialist and local services.

For families this was described as essential to enable them to understand their condition, know where to get support and make decisions about treatments.

It is hoped that our feedback will guide the Department of Health to reshape the Rare Disease Plan so that all future rare disease services will be designed in a way that better meets the needs of families.

Doctors raising awareness - news from the Bristol Barth Syndrome Service team

Dr. Colin Steward has commenced a series of lectures at the major paediatric hospitals throughout the UK. He has given a grand round presentation on Barth syndrome (BTHS) at Birmingham Children's Hospital and the Royal Liverpool Children's Hospital, Alder Hey. Further lectures are planned in Manchester and other northern cities. Dr. Beverly Tsai-Goodman will also give a lecture on BTHS at the British Congenital Cardiac Association in Belfast in November. These presentations are backed up by information available through the Service website and pamphlets about the disease from both the Service and Barth Syndrome Trust.

RAISING AWARENESS IN FRANCE

By Florence Mannes, Chair, Association Barth France

As Barth syndrome is quite unknown in France, there is a lot to be done in order to raise awareness among the general public, doctors, and also the affected families.

Raising awareness among the general public

As many members of Barth France are runners or triathletes, the easiest way for Barth France to raise awareness is to have as many people as possible participating in races wearing the Barth France colors. For example, 80 people with the Barth logo on their tee shirts, ran the Paris Half Marathon in March, and 60 ran the Paris Marathon in April. Barth France also attended the Paris Marathon Running Fair, distributing information leaflets during the three days of the fair (40,000 visitors). The local newspaper published articles about the participation of Barth France in these events.

Barth France athletes will participate in other races, triathlons and the Ironman during the coming summer and will also take part in the 10km "Swim across Paris" (swimming 6.2 miles in the River Seine). Each of our athletes receives information on the syndrome, so that they can explain what they are running for and raise awareness. If each of our marathon runners talks about Barth syndrome to five different persons, it means that 400 new people will be aware of Barth syndrome.

Raising awareness among doctors

Although some doctors we have met are fully aware of Barth syndrome, we have discovered that many doctors in France are not well informed about the disorder, and that, possibly, some children do not get the appropriate diagnosis, or medical follow-up.

(Cont'd on page 17)

(Cont'd from page 16)

By financially supporting a study of French Barth syndrome cases, we hope to be able to inform more doctors about this disorder and to identify new cases. With the support of professors who specialize in Cardiology, Hematology, Genetics, and Metabolism, Dr. Charlotte Rigaud will write a thesis on Barth syndrome to analyse the medical data of French Barth boys/men. In order to do so, the first step is to contact as many doctors as possible to determine if they have, among their patients, boys who might be affected by Barth syndrome. Once this is achieved, Dr. Rigaud will review the medical data of the Barth patients "recruited" in order to compare them.

Back in 2009, only one case of Barth syndrome had been identified through the French Severe Chronic Neutropenia Registry. As of today, the register records 14 cases, and, through this study, we hope that more French Barth boys will be diagnosed correctly.

Raising awareness among affected families

It seems that some of the French affected families are not really familiar with English and that they do not have access to the news and precious information shared on BSF's Listserv. On her way back from the Bristol clinic, Valérie, Jules' mother, had the idea of preparing a monthly summary in French of the posts sent to the Listserv, to send to the French speaking families.

We believe that raising awareness is also making sure that all the affected families have easy access to all the information available. We are also doing our best to translate the BSF Fact Sheets into French.

There is still a very long way to go to make sure each boy affected with Barth syndrome gets an appropriate diagnosis and treatment as early in life as possible, but we believe that each small step leads us to this goal, one step at a time.



Loyal members of France's Team Will with the Barth logo on their tee shirts participated in the Paris Marathon on April 15, 2012 in support of Barth France.
(Photo courtesy of Barth France ~ 2012)

BSF Thanks Our Donors the Bottom of Our Barth Hearts

By Stephen B. McCurdy, Chairman, Barth Syndrome Foundation

Twice a year, BSF publishes this newsletter and sends it to every family member, volunteer, scientist and clinician associated with Barth syndrome (whose address we have — please make sure that you keep us up to date on your current address!) and every donor who has sent us \$50 or more in the previous 18 months.

The Barth Syndrome Journal is one of several vehicles we use to try to stay close to the people who make up this global community and to keep you all informed about each other and BSF. Lynda Sedefian and her volunteer publications team (Nigel and Lorna Moore representing BSTrust, Les Morris representing BSFCa, and Kate McCurdy representing BSF) get regular accolades for the quality and content of the Journal, our web-site at www.barthsyndrome.org, our annual report, our various educational and awareness brochures and our Conference program materials — and they should! She and her team help us maintain our image and reputation as a small but professional advocacy group that continues to have an impact far larger than virtually any other rare disease affecting a comparable number of people.

I am telling you this for two reasons: 1) because Lynda and her team don't get enough credit for all that they do for us, and 2) to give you but one example of the broad and incredible critical range of activities supported by your donations!

In the year just finished — 2011, BSF raised just over \$700,000, and managed our budget closely to produce a net deficit of only \$35,000. Our largest program expenses are in Science and Medicine and include our annual research grant program and the Barth Registry & Repository (BRR) — our growing repository of DNA, cell lines, tissue samples and medical data contributed by our affected boys and young men. Every day (and often at night) our Family Services program provides information, insight, support and optimism for families struggling to cope with this disorder...and our communication program keeps everyone connected.

We do a lot with the resources you give to us. Our mighty staff and our volunteers stretch every nickel to get the most value for our investments. We want you to know that we appreciate every dollar...indeed every penny that you entrust to us so that we can all be proud — not just of what we accomplish, but of how we do it! Thank you from the bottom of our Barth hearts!

We've been busy since we last reported to you in our Fall 2011 Barth Syndrome Journal:



The late Paula & Woody Varner
(Photo courtesy of Sue Wilkins)

Sue and Mike Wilkins continued to add to the Paula and Woody Varner Fund, asking their friends and all those who admired and respected Sue's parents to support the science and medicine programs of BSF as Paula and Woody would have wanted them to. Among other things, the Varner Fund has funded the development of our Barth mouse — a many-year effort to create the first mammal with all the characteristics of Barth syndrome that can be studied by our scientists. They also fund the Varner Award, which will be awarded again at this year's Conference in June to a "Pioneer" who has made the greatest contribution to the advancement of science and greater understanding of Barth syndrome.

Tiffini Allen is quickly becoming a fund raising phenomenon at BSF! She created the "Hey, Hey Henry" event at the Chicago Cubs last year, and donations were still flowing in long after the snows of winter blanketed Wrigley Field. Not satisfied, Tiffini then created what she called "Henry Cards" (do you see the developing theme here?) — pictures of Henry or any Barth boy or young man that families could send to their friends, telling them about Barth syndrome and inviting them to support BSF with a

donation. **John Wilkins**, one of our young men affected by Barth syndrome and member of BSF's Board of Directors, loved the idea and asked Tiffini to design his own "Henry Card" which he then sent out to his many friends and family members to raise money for the upcoming Barth Conference. Simple, warm and effective, "Henry Cards" are the newest thing in fund raising! You can't be cool if you haven't sent them out and/or received one in the mail. Collect yours today (and be inspired to send BSF a donation)! You can read more about Tiffini and her "Henry Cards" on page 14.

As previously reported, for over a decade, **Kate and Steve McCurdy**, (see by-line above), lacking the creativity to create a "Henry Card," have been sending out year-end letters to friends and family with updates on BSF and their son, a 26 year old young man with Barth syndrome, and inviting donations to BSF. For over ten years, their friends have remained loyal

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John signing his "Henry Cards" to raise funds in support of BSF's 2012 Conference.

(Photo courtesy of BSF ~ 2012)

contributors to BSF, earning the undying gratitude of Kate and Steve and a growing host of Barth families. "We find that friends really want to help, but as is often the case, they don't always know how. Knowing how devoted we are to finding a cure for Barth, a donation to the Science and Medicine Fund is the obvious answer," say the McCurdys.

2012 Barth Conference donations. There are a number of opportunities to make a donation specifically in support of the June Barth Conference, but they are going fast! This Conference brings together families (including the largest gathering of affected individuals ever assembled to date), scientists, clinicians and young researchers for a five day period of clinical consultations, scientific presentations, educational programs and fellowships – all at the Don CeSar Resort in St. Pete Beach, Florida. As Lindsay Groff, BSF's Executive Director says, "This is actually at least four simultaneous conferences – for the parents and grandparents, for the affected boys and young men, for the siblings and for the Doctors and Scientists!" Sponsorship materials can be found on the BSF Website at: <http://www.barthsyndrome.org/english/View.asp?x=1688>. There will also be

an auction of valuable items that might appeal to a hard working doctor, scientist or family member such as gift certificates, sports/concert tickets, jewelry, cookbooks, spa/bath items, and handmade articles. If you would like to donate an item for the auction, please e-mail lindsay.groff@barthsyndrome.org.

Honoraria and Memorials. One of the most thoughtful ways to honor a special occasion like a wedding, a bar/bat mitzvah, or a birthday is to make a gift in the name of your friend to BSF. As long as we have the address of the honoree, BSF always lets them know of the gift made in their name. It's a wonderful tradition that honors everyone — the donor, the honoree and BSF! Likewise, BSF receives many gifts in fond memory of a loved one who has passed away. Barth syndrome affects everyone in a family, directly or indirectly: grandparents who finally understand why they might have lost a baby boy so many years ago; siblings who may discover they are carriers; parents who want to provide an equally loving and healthy environment for all their children, affected or not; aunts, uncles, cousins, in every generation are affected. Honoraria, memorials, and even legacy gifts in wills are all wonderful ways to honor a loved one and a special occasion with a gift to BSF in their name.

Late Breaking News – BSF Receives Conference Grants from the National Institutes of Health (NIH) Office of Rare Diseases Research (ORDR) and the National Heart, Lung and Blood Institute (NHLBI). Dr. Matt Toth, BSF's Science Director applied to both of these entities at the NIH for assistance in funding the scientific portion of the International Barth Syndrome Scientific, Medical and Family Conference to be held in June of 2012 and received a high enough score that both ORDR and NHLBI agreed to provide funding!

All of us at BSF are so grateful for the continued support of all of our many donors whose names are listed on the Power of Kindness pages in this Journal. Nothing we do could be done without your help and we are all so much better for your quiet presence in our little community. Thank You!

New Vice Chairman Appointed



Marc Sernel
(Photo courtesy of BSF ~ 2009)

I am delighted to announce that Marc Sernel, who has served on the Barth Syndrome Foundation (BSF) Board of Directors and as Corporate Secretary since early 2009, has agreed to serve as Vice Chairman for the Board beginning immediately. Marc and his wife Tracy have a daughter and two sons, one of whom has Barth syndrome. They joined BSF in 2006, and attended their first conference that year in Orlando. Marc has been a partner at the law firm of Kirkland & Ellis LLP in Chicago since 2003. With degrees in chemical engineering and law, he has been an especially valuable advisor and Board member as BSF engages more deeply in research and potential medical treatments, and his expertise keeps him moving constantly between the scientific and family sessions at the conferences. As he has three times previously, Marc will attend our Conference at the end of June, so please join me in thanking him for taking on this additional role when you see him next month.

Steve McCurdy, BSF Board Chairman

Barth Syndrome Trust

Sharing skills, sharing knowledge

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

In early 2012, volunteers from all over the UK met up in Hampshire. At this meeting, Dr. Vanessa Garratt and Debbie Riddiford from the Bristol Barth Syndrome Service introduced a draft education leaflet, and together we developed two new guides.

New Education leaflets for schools and families

The Education Booklet is almost ready for distribution, and we hope it will serve as an invaluable aid to families and teachers. We have also created a Short Guide for Teachers so that they can have all the most important information at their fingertips.

If you would like a free copy, please e-mail the Trust on info@barthsyndrome.org.uk.

Can you help?

We rely on unpaid volunteers and believe that everyone has a particular skill that can be used in order to help our boys and help us reach our goal. What can you do to help? Do you know someone who might join us in this very rewarding endeavour? If you would like to chat to someone about perhaps volunteering a little of your time, please contact us on info@barthsyndrome.org.uk.

We particularly need volunteers with any of the following skills or experience:

- Computer skills – Microsoft Access, Excel, publishing, graphic design;
- Organisational skills to help arrange meetings, workshops, focus groups and family days or help with administrative work;
- People skills and the ability to travel to raise awareness of Barth syndrome by talking about your experiences of Barth syndrome; join our Family Services team and be a lifeline for new families joining the Trust; help with fundraising;
- Writing or design to join our Publications team and help us with this newsletter or in creating new brochures and awareness material;
- Leadership skills, business skills and strategic planning. Why not apply to join the BST Board of Trustees?

NHS Bristol Barth Syndrome Clinic

The March clinic was well attended with patients coming from as far away as Scotland, Belgium and Switzerland. Feedback indicates that the clinics are seen as a focal point for providing quality care for patients and their families. Everyone gets the specialised and individual attention and information they need as well as the opportunity to mix with other families. At every clinic we ask, "What can we do better next time?" and with open and honest feedback from people attending, we continuously strive for excellence, involvement and patient-centred care.

(Cont'd on page 21)

Save the Date

Barth Syndrome NHS Clinic

Thursday 27th and Friday 28th September, 2012

Bristol Royal Hospital for Children

Followed by a Family Gathering on Saturday 29th September
Venue TBD

(Cont'd from page 20)



Benoit, age 12
(Photo courtesy of BStTrust ~ 2012)

Fun-filled family day

The place was buzzing as families competed in a friendly game of bowling at the local alley. Afterwards, everyone met at the Pizza Hut to enjoy a meal and a chat. The children enticed Dr. Steward into filling his bowl with as much ice cream as possible and made sure they had photographic evidence to pass on to Nicol Clayton, the Service dietician, in the hopes that she would tell him off and encourage him to replace his ice cream with either some vegetables or at least a tin of tuna fish (high in arginine!).

For Benoit, 12 years old from Switzerland pictured here, this was the first time he had met another person with Barth syndrome. For our Barth families, this was the time to be together. For all of us, it was an incredibly joyful experience to see young Joe – looking so well – after his heart transplant; a journey which took more than a year and which we followed every step of the way. (See article on page 15 for more details of Joe's story.)

Financial update for 2011

Income for the 2011 period was just under £20 000, with expenditure totalling approximately £30 000 of which £25 000 was for funding Dr. Anton I. De Kroon's research project: "**The preferred acyl chain donor of *Taz1p* in the acylation of monolysocardiolipin.**" We're very grateful to all our donors and fundraisers and would urge you to please consider making a donation or organising a fundraising event for us this year as we urgently need more funds to continue our work.

Update on the NHS Bristol Barth Syndrome Service

By Colin G. Steward, FRCP, FRCPCH, PhD, Clinical Lead, NHS Bristol Barth Syndrome Clinic

During the past year, we have received specimens for testing principally from geneticists and pathologists together with cardiologists from Bristol and several London teaching hospitals. Although requests for testing are increasing, we are still concerned about the lack of routine testing from cardiac departments, especially from areas outside southern England.

We are currently running two clinics per year for UK patients as well as seeing newly diagnosed children for ad hoc assessment/discussions. We plan to develop a transitional adult clinic in 2012 to be led by Dr. Rob Martin (Consultant Cardiologist) and held in the Bristol Heart Institute as a more appropriate site for transitional adult care.

We would like to look at involving young people more in the following year to help look at how the Service could support them better. In the March clinic, a boys' groups was held by a youth worker. The boys and their families have been asked about whether this is a useful aspect of the Service and whether it should be repeated. Dr. Vanessa Garratt, Clinical Psychologist to the Service, has been asked to host a number of workshops for families at the forthcoming BSF Conference in Florida in June 2012 which should further inform this process.



Families at the Bristol Clinic, March 2012
(Photo courtesy of BStTrust ~ 2012)

Over the last year, a focus group was held with families to look at a draft educational brochure. Following the clinic in March 2012, a bereavement focus group was held to give bereaved families and families with living children an opportunity to talk about what services they would like to support parents and siblings. A number of action points were generated from this meeting. Some families have volunteered to help take these forward and be involved in developing resources.

Patient satisfaction

Patient satisfaction questionnaires continue to provide useful feedback for the team to improve future clinics. Feedback from the clinics in 2011 and 2012 has been very positive, in particular that the clinics and Service provide opportunities to learn more about Barth syndrome and to talk to all the different professionals.

Barth Syndrome Trust

Sharing skills, sharing knowledge

(Cont'd from page 21)

However, some concerns have been raised in the recent clinic about how tiring the multiple appointments can be for this group of patients who struggle particularly with fatigue. The team are looking at this with the Barth Syndrome Trust and are keen to explore the potential of telemedicine to reduce the burden of appointments required at any one clinic.

Future plans

- Develop patient-held records.
- Develop resources to support families who are worried about losing a child and bereaved families.
- Trial the use of the new educational brochure in some schools.
- Offer further dedicated separate sessions for boys and families within the Clinic.
- Trial telemedicine appointments in some areas to reduce overall Clinic burden.
- Continue to push for incorporation of cardiolipin testing into standard protocols for investigation of cardiomyopathy.
- Offer overnight oxygen and carbon dioxide monitoring to patients who have problems with lethargy/fatigue as we are becoming aware that sleep apnoeas can be a significant problem in this disease.
- Develop an instruction video, preferably in the form of animation for reasons of child friendliness and palatability, on how to perform home fingerprick blood count testing, important for evaluation of neutropenia and G-CSF responses.

We continue to work closely with the Barth Syndrome Trust to design and improve the Service. The Trust has also kindly funded the part-time neuropsychological assessment post for a year.

Note from BST Chair: When people ask us “What is Barth syndrome?”, we often explain it with a quick list of symptoms, saying that it affects the heart, muscles, immune system and growth. However, for those of us living with it, we know that it can sometimes affect everything. There is the medical side, much of which is still unexplained but there are also the day-to-day challenges of struggling with a condition that can cause exhaustion and difficulties in schooling and friendships and independence. We never really know what’s around the corner and that can be frightening at times.

Through our worldwide community, families can support each other which makes the journey so much easier. And after reading about all the work being done on our behalf by the Bristol team, we are doubly grateful for their pro-active and patient-centred approach which has drawn them into the heart of our larger Barth family.



(L-R) Emilie, Alejandro, Joe and Aneira at the Clinic.
(Photo courtesy of BSTrust ~ 2012)



(L-R) Will and Oliver enjoy table tennis during the Clinic.
(Photo courtesy of BSTrust ~ 2012)

Barth Syndrome Trust

From Pennies to Pounds

Despite the recession, the Barth Syndrome Trust's (BST) families, volunteers and friends have been raising much needed funds through donations, hard work and innovative ideas. Very many thanks to everyone for your efforts and generosity.

Donations: We have received donations from the UK, Europe and Australia. HSBC in Bristol gave another donation, £215 in memory of baby Jack. Watson Marlow Company and Staff donated £100 in lieu of sending Christmas cards. The Bat and Ball Club table tennis tournaments have for many years levied 'fines' and this year £275 was collected. The Basingstoke Tennis Club Quiz winners led by Colin Phillimore, again selected BST to receive the night's profits – £250.

Yorkshire and Derbyshire fundraisers: Friends and colleagues of Julie Woolley and her mother Linda Barratt have been supportive as always. Wilf and Brenda Smith's raffle raised £200. From Derbyshire Dales District Council: £77 – Payroll Office's Groovy 1970's Grotto; £259 – Revenues Department's book sale and raffle; £15 from the Benefits section in lieu of sending Christmas cards. Stannington Friendship Agewell Group in Sheffield donated £200.



Hearts for Barth

In memory of Philip Brown (22/05/2005 - 29/09/2009)

Hearts for Barth: Westgate Primary School, Otley, ran a 'Hearts for Barth' for Valentine's day so that pupils could buy Valentine hearts for their parents. Everybody pitched in to create beautiful hearts: mums, neighbours, grandparents. An amazing £292 was raised – thank you all. We are very grateful that the school still remembers Philip.

Claire Clements, who was Philip's childminder, gave BST £267 from the sale of books and toys.

Greetings from Wales: Isabel Easterbrook reports how a BST workshop inspired her to do some fundraising and raise awareness. Family, friends, and even strangers helped. Her sister, who owns a small cake business, made cupcakes to sell on Rare Disease Day, February 29th. Cake sales with awareness displays were held at the Royal Gwent Hospital, Legal & General, HMRC Cardiff and City & Guilds raising a total of £589. Thank you to all who helped and bought cakes and raffle tickets, especially to baker, Kate Riseborough, and the Legal & General for a matching donation.



Baking for Barth in Wales!

Thank you to: Caerphilly Masons for £107 from their quiz night; Rev. Dr. Sarah Rogers, Helen Murphy, Daphne Holdsworth and Gaynor Rees of St. Catherine's Church, Caerphilly for £106 from their bingo afternoon; and Paul and Margaret Challenger who invited Ralph Easterbrook to make a speech at a spiritualist evening and donated the proceeds of £342.

Sponsored events: Two fundraisers were inspired by young Mitchell. Sponsorship for Karen Olden's 2hr 15 min half marathon in Cambridge totalled over £550. Patricia Jones completed the gruelling London Marathon on 22nd April. Even an injury to her hip 14 miles in didn't stop her from reaching the end. Patricia raised over £1500.

In spite of fracturing his wrist and preparing for exams, Eden, brother of Jack who has Barth syndrome, has now 'reached France' having swum a distance equivalent to crossing the Channel to collect over £1000! If you would like to support these brave efforts, donation pages are still open on Bmycharity.com.

Forthcoming events: Lizzie Stagg and partner Kevin will run the Bupa London 10,000 on 27th May in memory of baby Oscar. Joshua Haycock and friends will cycle 900 miles from John O'Groats to Land's End. Joshua writes, "Planning and training for this is the most difficult thing I've ever had to do, but knowing that it's for a great cause helps push me to my absolute maximum. It will be worth it, for my nephew and BST." See Bmycharity.com. Scott, 11, friend of Nick who has Barth syndrome, is planning to attempt the 3 Peaks Challenge, climbing the highest peaks in Scotland, England and Wales, all in 24 hours. (www.sponsor-me.org/3peaks2012)

Barth Syndrome Foundation of Canada

President's Report — Spring 2012 Newsletter

By Lynn Elwood, President, Barth Syndrome Foundation of Canada

Looking back on the accomplishments of 2011 and early 2012, there is a lot that has been done within Canada. We have held a number of successful fundraisers including the Golf Tournament, and a number of individual events such as Boogie for Barth, a 50th Birthday bash and a Poinsettia sale. We have received a number of donations and expanded the amount of grant money we have received, partially through ties from the US BSF organization. This, coupled with prudent expense management by our Board and Executive, resulted in a strong financial position for 2011.

SUMMARY OF FINANCIALS	
Statement of Operations (Year Ending December 31, 2011)	
Revenue	\$66,854
Expenses	\$26,759
Excess of Revenue over Expenses before Grant Funding	\$40,095
Research Grant Funding	\$38,234
Excess of Revenue over Expenses, Funding	\$ 1,861
Statement of Changes in Fund Balances (Year Ending December 31, 2011)	
Opening Balance	\$89,448
Excess of Revenue over expenses & funding	\$ 1,861
Closing Balance	\$91,309

The Scientific and Medical Grant funded by BSF of Canada last year was, "**A screen for drug leads for the treatment of Barth syndrome**," submitted by Christopher R. McMaster, PhD, Professor of Pediatrics and Biochemistry, Dalhousie University, Halifax, Canada. This year, we have once again been able to fund a complete grant, "**MLCL AT-1 elevates cardioliipin and mitochondrial function in cardiac myocytes of *taz* knockdown mice**," by Grant Hatch, PhD, Professor, University of Manitoba, Winnipeg, Manitoba, Canada.

We are pleased to be able to fund BSF's international Scientific & Medical Advisory Board's recommended grants for these Canadian researchers and very much appreciate the closer working relationship that is developing between them and the Canadian organization.

We have had some good success in our Family Services program this year, with members of our Board being in touch with and having the opportunity to meet with almost all of our boys and men affected by Barth syndrome. In April, we were able to bring five of them together in a family outreach event that accompanied our Annual General Meeting. A number of the men affected with Barth syndrome and their siblings are now official voting members of BSF of Canada, and it is a pleasure to have more of them participating in the AGM meeting itself.

We continue to publish Canadian newsletters (now with new team of Les Morris and Paula Sisson) and those are well received by our family and donor population. This is one form of awareness in our region, and another significant project this year was the trip out east where there were both awareness and Family Outreach events. While we have a number of outreach methods, opportunities like we had in Halifax with Dr. McMaster and several doctors are some of the most beneficial, as we are able to meet in person with a large number of physicians and scientists.

Volunteers are what make this organization strong. This year has presented some challenges for some of our regular volunteers, and people who were previously able to spend more time helping had less of their time to share. However, the depth and passion of our volunteer group, including the Board and Executive, has accomplished great things throughout the year resulting in ambitious plans for the coming year.

Canadian Annual General Meeting Combines Business and Bowling

By Chris Hope, BSFCa Treasurer and Susan Hone, BSFCa Secretary

We are looking forward to the upcoming BSF International Conference. The Canadian volunteers are an important part of the organizing team and volunteers at the Conference, and it is a great opportunity for the international community to meet and work together. We have been fortunate to sponsor some portions of the Conference such as the Scientific Poster Session, Youth Activities, and funding of some researcher travel expenses. We are looking forward to seeing everyone there and to watching our boys and men bond together and grow as they do at every conference.

The Barth Syndrome Foundation of Canada's Annual General Meeting (AGM) was held on April 21, 2011 for all members. In order to give everyone a chance to take part, members are given the option of attending this either by phone, or in person, in Toronto, ON at the fantastic office facilities arranged for us by Ian Morris.

The purpose of this yearly meeting is for the Board and Executive to give members an assessment of the previous year's activities, to review the financial statements, and to give an overview of the current year's plans and events, along with a projected budget.

This year, we had five individuals with Barth syndrome in attendance—Jared and his family once again made the trek from Regina, SK to Toronto, ON. Due to being temporarily non-ambulatory, our newsletter editor, Les Morris, was unable to attend in person, but he continued to give his valuable input via the phone—it just wouldn't be a BSFCa AGM without Les' contributions.

This once-a-year event is a great occasion to reflect on the work that has been accomplished and to acknowledge the volunteer hours and effort that everyone has contributed to ensure that our programs run smoothly. Special plaques were presented this year to Lois Galbraith and Cathy Ritter for their exceptional hard work and dedication. They are both essential to the function of our Foundation.

After the business portion of the day, it was on to the social time. After a bit of catching-up and friendly chit chat while munching on some goodies, it was on to the bowling alley. Everyone was eager to show their stuff, and balls quickly started flying in all directions. A fun time was had by all, and we are looking forward to next year.



(Photos courtesy of BSFCa ~ 2011 & 2012)

Paris Marathon—En masse for Barth France

By Florence Mannes, President, Association Barth France



(L-R) Arnaud and Adrien, together with Philippe and Raph as they cross the finish line!
(Photo courtesy of Barth France ~ 2012)

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

It all started last October, after the Brussels half marathon ... five of us decided to run the Paris Marathon, wearing Barth tee-shirts ... Pierre and Philippe, who would run their 8th and 9th marathons, Marc and Arnaud (for the 3rd time), and I, for my first time. ... The type of challenge you sign up for in the evening and when you wake up the next morning you wonder why you said yes.

Our friends, relatives, colleagues responded and Philippe's friend, who is a former triathlon champion, said he would come from southern France with his team, Pierre's college friends said they would follow, Arnaud and Marc's colleagues as well, and even our Hawaiian friend agreed to cross half the planet to come and wear the Barth colors in Paris!... Within two months, we were 80 planning to run the Paris Half Marathon, and 60 the Paris Marathon. ... This was totally unexpected, but such great news!

We then designed, negotiated prices and ordered the tee shirts ... which were not delivered to the right address, not compliant with what we had ordered, ... and after sending all of them to the future runners, we realized that we were running short of tee shirts...so reorder, wrong delivery again, ... but, finally, everyone managed to get his Barth tee-shirt before the race.

There we were, six weeks before the Marathon...and Pierre managed to contact the Paris Marathon organization team, and they agreed to grant Barth France an exhibition stand at the Running Fair held for three days before the Marathon, with 40,000 visitors, as well as a music stand along the route.

This was such great news ... but SO unexpected ! We had NEVER done that before, and had no idea of how to organize the fair stand, nor the race-side stand ... then once again, we called our friends ... to get ideas for products to be sold on the stand, to design information leaflets and banners and have them printed (at no cost ... because these are real friends!), to play music along the route, to rent the music sound system, to have articles published in the press. We also needed volunteers to man the stand for the three days of the fair, to distribute leaflets, sell tee shirts and bracelets, ... some of them took days off to do so!

Last but not least, we needed someone to play music during the race, and to be in the center of Paris at 7 am on Sunday morning to install the stand, the sound system, the information banner...and to stay there for six hours ... the problem was: as ALL our friends, and many friends of our friends were running the Marathon, it was hard to find someone who would be available to man the stand (we did not think about that at first!). ... Raph's grandfather was, finally, designated to be responsible for the race stand.

On Friday night, two days before the race, when I received a phone call from the band we had found, saying they would not be able to play during the race as planned, we were about to give up...running a marathon is one thing, organizing everything for a charity to be visible during a fair and a 40,000 person race is something totally different. ... While picking up my kids on Friday I told a mother about our problems and she took out her phone. In less than 15 minutes, she had recruited her son to be a DJ at the race, had contacted five different bands to see if they would be available on the next Sunday, and re-organized her schedule so that she could be at the stand during the whole race. I was so thrilled at such commitment from someone I did not know that well.

Sunday morning, the rendezvous was at our house at 6.45 am ... and it was so strange to see all these people, some of whom I had never even met before, in our living room (which was more of a changing room at that time!), wearing Barth tee shirts! and it was so great to have almost everyone in the photo taken in front of the Arc de Triomphe just before the race!

42.2 km is a long way....but seeing Raph at 10K was amazing, and gave us the strength to run to the finish line. Running for all the Barth boys, to raise awareness, and taking into account that Raph would probably never be able to do so made the experience special, and helped us when it became harder.

42.2 km is a long way, and we would have never achieved this without the support of our friends and families.

(Cont'd on page 28)

60 Marathonien(ne)s pour Barth!

By Florence Mannes, Présidente de l'association Barth France



(L-R) Arnaud and Adrien, together with Philippe and Raph as they cross the finish line!
(Photo courtesy of Barth France ~ 2012)

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

Tout a commencé en octobre dernier, au lendemain du Semi Marathon de Bruxelles... Nous étions 5, et avons pris la décision de courir le prochain marathon de Paris sous les couleurs de Barth France... Il y avait Pierre et Philippe, qui prendraient, respectivement, le départ d'un marathon pour la 8ème et 9ème fois, Marc et Arnaud (qui s'engageaient ainsi pour leur 3ème marathon), et moi, pour la toute première fois! C'est le type de défi que l'on relève, un soir, et dont on se demande, au réveil le lendemain, si on n'a pas fait une bêtise...

Mais nos amis, nos proches, nos collègues ont tous répondu présents, et nombreux d'entre eux ce sont inscrits au Marathon de Paris à nos côtés. Il y a eu Eric, ami de toujours de la famille Mannes, ancien champion de triathlon, qui s'est engagé à venir, du sud de la France, avec son équipe, il y a eu les amis d'école de Pierre, les collègues de Marc et d'Arnaud, et même notre pote d'Hawaii qui a accepté de traverser la moitié de la planète pour venir courir à Paris sous les couleurs de Barth France....

Ainsi, et en moins de 2 mois, nous étions 80 inscrits au Semi Marathon de Paris, et 60 au Marathon. Nous ne nous attendions pas à un tel soutien, mais nous étions ravis de voir tout ce monde soutenir notre projet!

Ensuite, il a fallu commander les tee-shirts, en négocier le prix... pour découvrir in fine, et à quelques jours du Semi Marathon que la livraison, qui n'avait pas été faite au bon endroit, n'était pas conforme à la commande... Ensuite, il a fallu les faire parvenir à tous les membres de l'équipe... Pour réaliser, que, nous allions manquer de tee-shirts... donc nouvelle commande, nouvelle livraison (mauvaise adresse encore!)... mais, finalement, tout le monde a pu avoir son tee-shirt avant la course (non sans mal!).

Et nous voici, 6 semaines avant le marathon... Pierre nous appelle pour nous annoncer qu'il a réussi à obtenir un stand sur le running expo, la foire dédiée à la course à pieds, qui se tient, pendant 3 jours, avant le Marathon, et qui voit passer plus de 40.000 personnes, ainsi qu'un stand lors du Marathon, sur le bord de la route.

Une fois encore, une très bonne nouvelle... Mais très inattendue! Nous n'avions jamais participé à ce genre d'évènement par le passé, et n'avions pas la moindre idée de comment organiser un stand, ou de la manière dont il fallait être présents lors de la course... Du coup, une fois encore, on a du faire appel aux copains... pour les bonnes idées des bracelets en silicone, pour jouer de la musique le long de la course, pour la location de la sono, pour la publication d'articles dans la presse. Il nous fallait aussi des volontaires pour tenir, avec nous, et durant 3 jours, le stand du Running expo, pour distribuer des prospectus d'information sur le Syndrome, pour vendre des bracelets et des tee-shirts... certains de nos amis ont même pris une journée de vacances pour ça!

Enfin, il nous fallait trouver quelqu'un pour jouer de la musique sur le stand le long du Marathon, et quelqu'un de confiance qui serait prêt à être en plein Paris, un dimanche matin à 7 heures, pour installer le stand, la sono, les affiches... et pour y rester 6 heures d'affilée... notre seul problème: TOUS nos amis, et beaucoup des amis de nos amis, courraient le Marathon... et il nous fallait pourtant trouver quelqu'un pour tenir le stand (nous n'avions pas pensé à ce type de problème au départ!). Finalement, nous avons « recruté » le Papou de Raph...

Vendredi, deux jours avant la course, je reçois un coup de téléphone du groupe de musique qui était censé assurer l'animation musicale pendant le marathon... Pour me dire qu'ils ne pourraient pas être présents... et il est de ces moments où vous avez envie de tout laisser tomber... courir 42km est une chose, faire le maximum pour qu'une association soit convenablement représentée pendant une foire, et lors d'un marathon en est une autre... C'est ce que j'explique à une maman de l'école de Raph et de ses frères, le vendredi soir... En moins de 15 minutes, son fils était d'accord pour assurer le rôle de DJ pendant le marathon, elle avait contacté pas moins de 5 groupes de musiques différents, et modifié le planning de tout son week-end pour être sûre de pouvoir être présente sur le stand lors du Marathon... J'ai été stupéfaite d'un tel dévouement, de la part de quelqu'un que je ne connaissais que depuis quelques semaines.

Dimanche matin, le grand jour, le rendez vous est donné à la maison à 6h45... étrange de voir tout ce monde, les copains mais aussi tous ceux que je n'avais jamais vus, dans notre salon (qui ressemblait plus, pour l'occasion, à un vestiaire), porter le même tee-shirt Barth France... et c'était fantastique de voir tout le monde présent pour la photo, devant l'Arc de Triomphe, quelques minutes avant la course!

(Suite à la page 29)

Paris Marathon—En masse for Barth France

(Cont'd from page 26)

42.2 km is a long way, but the search for a cure for Barth syndrome is also a long journey....and we all hope that one day we can say, with the same smile that Philippe had when he crossed the Paris Marathon finish line with our son Raph on his shoulders ... we defeated Barth syndrome as we defeated the 42.2 km.

A small association like ours is really a matter of friends and family, and none of this could have been achieved without their help, love and support!!



Photo courtesy of Barth France ~ 2012

Ironman4Barth ... Swimming, Biking, Running, for Barth Boys

Ironman4barth is a team of triathletes who are running triathlon, half ironman or ironman in order to raise funds and awareness for Barth syndrome. Inspired by Gary Rodbell's successful experience with Team Will, they were, in 2011, seven men, seven dads, who decided to run the France Ironman ... and as they loved it, they have put on their agendas, in addition to the Paris Half Marathon and Marathon, some new challenges for 2012....

- Nice France Ironman, again
- Alpes d'Huez Triathlon
- Half Ironman of Aix les Bain (with three women as a relay team)
- Embrunman (Ironman distance)
- 10 km swimming in the River Seine
- Paris Triathlon
- 20km of Paris

In 2011, a total of 2,200 km were run by a dozen athletes wearing the Barth shirt. For 2012, our target is to run more than 7,000 km as a whole wearing the Barth tee shirts. After seeing us in races, some people contacted us to run for Barth syndrome; it is great to know that people who have no relation with Barth syndrome now run for our cause, and we hope to have more athletes in our colors each year.

A French Doctor Preparing a Thesis on Barth Syndrome

One year ago, Barth France attended a triathlon, and we distributed information leaflets on Barth syndrome. It happens that we gave one to a doctor who is one of the best professors in Hematology in France...He seemed to be interested in learning more about Barth syndrome....On meeting his team, we discovered that a few years previously they had created a national register for neutropenia, and that, within all the neutropenias, they had identified Barth syndrome, and recorded one French case in 2009, and 14 by the end of 2011.

Dr. Donadiou, who is responsible for the Neutropenia Registry, introduced us to Dr. Charlotte Rigaud, who is a French doctor, and Barth France agreed to financially support her in preparing her thesis on Barth syndrome. The goal of her thesis is to systematically analyze the medical data of Barth patients, in order to find out if there might be common factors that would potentially explain the ups and downs of Barth syndrome.

As we anticipate that 14 cases do not represent all the French cases, the first step of the thesis is to contact all of the French hospitals, with the help of the members of the steering committee of the project, who all are well known specialists (cardiologists, hematologists, geneticians, and metabolists) to see if they have other possible Barth cases not known to us.

The second step would be to compare all the medical data in a formalized way, and to analyze them to better understand the mechanisms of the syndrome.

Our next goal, through this study, and with the support of the members of the steering committee, would be to organize a Family gathering, together with clinics, in Paris, for all Barth syndrome patients willing to attend, allowing them to meet doctors who are familiar with the disorder and who have met other Barth patients.

Dr. Charlotte Rigaud and Chris Ottolenghi (biologist, member of the steering committee), will attend BSF 2012 Conference, with the support of Barth France.

60 Marathonienens pour Barth!

(Suite de la page 27)

42,2 km, c'est long, mais le chemin vers un traitement pour le Syndrome de Barth est également un très long périple, et nous espérons sincèrement pouvoir, un jour, dire, avec le même sourire que celui de Phil passant la ligne d'arrivée du marathon avec Raph sur ses épaules, que nous avons vaincu le Syndrome de Barth, comme nous avons vaincu les 42,2 km.

Une petite association comme Barth France repose, avant tout, sur les amis et la famille, et rien de tout cela n'aurait pu être réalisé sans leur aide, leur soutien et leur amour.



Photo courtesy of Barth France ~ 2012

Ironman4Barth ... Nager, Rouler, Courir, pour Barth

Ironman4barth, c'est une équipe de triathlètes qui participe à des triathlons, semi Ironman, Ironman dans le but de faire connaître le syndrome de Barth, et de financer la recherche sur le Syndrome. Avec comme exemple Gary Rodbell, à l'initiative de la création d'une superbe équipe de triathlètes pour la Barth Syndrome Foundation, aux USA, ils étaient 7, en 2011, à s'inscrire sur l'Ironman France, à Nice...et, parce qu'ils ont aimé cela, ils ont ajouté d'autres défis à leur planning pour 2012:

- Ironman France, à Nice, une nouvelle fois
- Le Triathlon de l'Alpe d'Huez
- Le Semi Ironman d'Aix les Bains (avec une équipe féminine en relais)
- L'Embrunman (distance Ironman)
- Paris à la Nage (10km dans la Seine)
- Le Triathlon de Paris
- Les 20 km de Paris

En 2011, plus de 2.200 km ont été parcourus avec les couleurs de Barth France, par une douzaine de coureurs. En 2012, l'objectif est de parcourir plus de 7.000 km au total. Parce qu'ils ont vu nos tee-shirts lors des différentes courses, des sportifs nous ont contactés, pour courir sous les couleurs de Barth. C'est fantastique de savoir que des personnes qui n'ont aucune relation directe avec le Syndrome courent maintenant pour Barth, en espérant que notre équipe va continuer à grandir.

Une thèse sur le Syndrome de Barth

Dr. Donadieu, le responsable de ce registre, nous a présenté le Dr. Charlotte Rigaud, interne en pédiatrie, et Barth France a décidé de financer les recherches qu'elle ferait sur le Syndrome de Barth, dans le cadre de sa thèse. L'objectif de cette étude est, via l'analyse systématique des données médicales propres à chaque patient atteint du Syndrome de Barth, de déterminer s'il est possible d'expliquer les évolutions souvent inattendues de la maladie.

Dans la mesure où nous sommes conscients que les 14 cas recensés par le registre national des neutropénies ne représente pas l'ensemble des cas Barth en France, la première étape de cette étude est de prendre contact avec l'ensemble des hôpitaux français pour déterminer s'il existe d'autres cas, avérés ou suspectés, de Syndrome de Barth, dont nous n'aurions pas connaissance. Cette démarche est faite avec l'appui des membres du comité de pilotage de l'étude, qui sont tous des professeurs respectés dans leur spécialité (cardiologues, hématologues, généticiens, métabolistes).

La deuxième étape de l'étude sera l'analyse systématique des données médicales, pour avoir une meilleure compréhension des mécanismes de la maladie.

La prochaine étape, grâce à cette étude, et avec le soutien des membres du comité de pilotage, serait d'organiser une réunion des familles francophones, en proposant des consultations médicales avec des médecins qui connaissent bien le Syndrome de Barth, et qui suivent déjà d'autres patients.

Dr. Charlotte Rigaud et Dr. Chris Ottolenghi (biologiste, membre du comité de pilotage) seront présents lors de la BSF conférence de cet été.

Have You Moved Lately? Please Help Us Keep Your Information Current

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

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

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



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Power of Kindness

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 Thompson, Kim
 Thorne, Antony
 Thornes Farm Shop, Crediton
 Timerick, Jo
 Toon, Kay
 Towle, Teresa
 Tsai-Goodman, Dr. Bev
 Vecchi, Guilia & Antonio
 Walsh, Yvonne
 Wanders, Dr Ronald
 Ward, Anne & Barry
 Warner, Rosemary
 Watson-Marlow Limited
 Wessels, Leigh-Anne
 West, Laura
 Western, Sue
 Westgate Primary School, Otley
 White, Jan
 White, Mark
 Whiterod, Brenda
 Wilks, Gemma & Phil
 Williams, Mary
 Wilson, Samantha
 Wites, Agnieszka & Wojciech
 Witzani, Horst & Linde
 Woodward, Tracy & Ian
 Woollen, Kate
 Woollen, Rob
 Woolley, Scott & Julie



Jules (BTHS, age 3) enjoys his ride!
 (Photo courtesy of BSTRust~ 2012)



Eden with his brother,
 Jack (BTHS, age 5)
 (Photo courtesy of BSTRust~ 2011)



Alex (BTHS, age 11) with his brother,
 Ieuan (BTHS, age 14)
 (Photo courtesy of BSTRust~ 2012)

Power of Kindness



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 Aitken, Stewart
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 Basilio, Anita
 Basilio, Paul
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 Baxter, Katherine
 Baxter, Russ
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 Best, Rance
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 Bond, Darryl
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 Bonneau, Rodger - Bonneau Mechanical Services
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 Landscapes Inc.
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 Carter, Paul - Adapt Aviation Inc.
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 Cattelano, Valerio
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 Clarke, Mel
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 Coelho, Roberta

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 Country Meat Cuts
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 Daley, Sheldon
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 Davison, Marilyn
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 McJannett, Bob
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 Wright, Cameron
 Wright, Jessica
 Wuthrich, Grace
 Young, Jim
 Young, Lenora
 Young, Ron
 Zakarow, Bob
 Zavitz, Peter



Barth Syndrome Foundation

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Website: www.barthsyndrome.org



Wyatt (age 4)



Jack (age 19)

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