Barth Syndrome Foundation

Barth Syndrome Foundation

www.barthsyndrome.org

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

2010 Annual Report

Barth Syndrome Foundation 2010 Annual Report

In the life of a child, ten years can seem like a lifetime. For children with a rare metabolic disease like Barth syndrome it can be. Our job is to fix that.

CHAIRMAN'S LETTER

Ten years ago, parents of boys with Barth syndrome struggled to keep their sons alive. They were doing so alone, with little help from medical science and often very little guidance from their physicians. By 2001, papers had been published by Dr. Peter Barth and Dr. Richard Kelley that named the disorder, established its genetic origins and noted that its primary symptoms were cardiomyopathy, neutropenia and general muscle weakness. The papers also noted that Barth syndrome was often fatal.

The Barth Syndrome Foundation was created by a small group of parents who understood firsthand the hopelessness of facing this disease alone. They needed each other to share information and support. They needed their sons' doctors to learn from each other how to diagnose and treat Barth syndrome. And they needed the scientific community to study this disorder to better understand its underlying causes and develop better treatments and someday a cure. And as is always the case, they needed money to fund it all. But above all, they knew that none of this would happen if they did not take the lead themselves. And so the Barth Syndrome Foundation was created.

Ten years later, much has changed, for which we are all very grateful. And yet some things remain the same. 2010 was a year full of progress. And a time full of wrenching sadness too, as we lost four boys/young men in 2009 and two in 2010 to Barth syndrome.

The Barth Syndrome Scientific, Medical and Family Conference

Your Foundation held its Fifth International Scientific, Medical and Family Conference during the last week in July at the Renaissance at SeaWorld hotel in Orlando, Florida. The Conference opened with two days of clinics providing clinicians the opportunity to examine and take the histories of more Barth syndrome boys and young men than any of them will ever see in their careers. This clinic serves at least two purposes: (1) it allows the efficient collection of physiological data and historical medical information from patients with this rare disease; and (2) it provides opportunities for patients and patient family members to meet with physicians who have a substantial experience in treating BTHS individuals. In 2010, six distinct IRB-approved protocols were participated in by many of the BTHS individuals who attended the clinic. Most of the data collected are expected to lead to publications and/or ultimately to be available through the Barth Syndrome Registry and Repository (BRR) which is open to all interested researchers.

Next, over 65 scientists, physicians, and healthcare professionals then gathered to hear 26 scientists review their latest findings and to discuss the progress in Barth syndrome (BTHS) research and how it may lead to better treatments. In a separate but parallel set of meetings, nearly 50 BTHS individuals with their families also met to discuss issues of specific importance to their situation. This latter meeting was the largest single gathering of individuals affected by Barth syndrome the world has ever seen. In total, nearly 350 people attended this dual-track conference. The informal mixing of this diverse group of individuals at common meals, at the Poster Session and at the social function, now a traditional part of this conference series, creates a strong sense of community and strong personal dedication to a mutual cause.

BSF Science and Medical Programs

BSF started out the year by selecting seven highly qualified research grants from the largest number of applicants previously received by BSF, awarding a total of almost $280,000. BSF's affiliate – BSF of Canada – provided funding for one of the grants awarded to a scientist in Canada. BSF's affiliate – Barth Syndrome Trust (UK & Europe) – provided funding for one of the grants awarded to a scientist in The Netherlands. BSF and its affiliates have now funded $2 Million in research grants of up to $40,000 each, spawning over 55 peer-reviewed scientific and medical articles published with the support of BSF.

One of the most exciting announcements made in 2010 was that a mammalian model of Barth syndrome – a Barth mouse – had been created with funding by BSF. This mouse and its progeny have been shown to exhibit virtually all of the genetic and clinical manifestations of Barth syndrome and are already being used to advance valuable research including several of the research grants awarded recently in the 2010 grant cycle.

The other major breakthrough was accomplished by one of our affiliates – The Barth Syndrome Trust (BST) in the UK. Through the determined efforts of Michaela Damin, BST's President and a BSF Board member and Dr. Colin Steward of the Bristol Royal Hospital for Children, the UK's National Health Services funded the creation of the first comprehensive, multi-disciplinary clinic dedicated to Barth syndrome. This grant runs for five years and will serve affected individuals and their families throughout the UK and Europe. The Clinic will provide significantly improved care for children suffering from this rare disorder, greatly increase the exposure, experience and learning of the clinicians caring for this large group, and produce detailed medical histories and data over a five-year period for the Barth Syndrome Registry and Repository (BRR)… a triple play in American baseball (not sure what this would be in Cricket!)

You can read more about our accomplishments in Science and Medicine in our Science Director, Dr. Matt Toth's report, which follows.

BSF's Family Services and Awareness Programs

BSF's Family Services team continues to find and support our growing list of affected families through the Barth Syndrome Conference, our internet based listserv and regional outreach gatherings. The Family Services team is also available by phone 24x7 whenever a child is sick, in the emergency room or intensive care unit to provide connections to the Barth syndrome medical experts and comfort and reassurance to distraught families. For a small, geographically dispersed group, stressed to the breaking point by a serious disease, a caring community can be a real lifeline. The alternative is to be totally, deeply and irretrievably alone. Year after year, BSF Family Services cements a community that counts its members closer than friends and family. The programs create the opportunity to communicate, cooperate and provide mutual support. The character of BSF is shaped more by this team, led by Shelley Bowen, than by any other single program or activity… and is the primary reason we are as healthy an organization as any truly rare disease group in the world. Nothing else would sustain us through the loss of five of our sons to Barth syndrome over the past 15 months, including Shelley's own son, Michael Bowen.

Shelley's report on Family Services and Awareness provides many more details on 2010 accomplishments.

BSF Governance and Administration

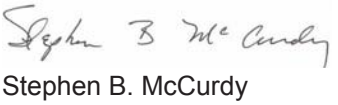
The BSF Board takes its governance responsibilities seriously. In 2010, BSF remained fully compliant with a host of new standards set by the IRS, the Better Business Bureau and National Health Council – all groups whose review and approval BSF has always received and valued. In addition, the Board created its first policy handbook covering its employees, their benefits and our mutual expectations of each other.

We are also concerned that the Board continue to attract new, dedicated, highly qualified members. Our by-laws require that the majority of Board members be directly related to someone affected by Barth syndrome. In addition, our by-laws limit the number of consecutive terms a Board member can serve to two three-year terms following the adoption of this rule. Several of our founding Board members must begin to step down from the Board beginning in 2013 including our Chairman, Steve McCurdy and Michaela Damin from BST – UK. In light of this, we have recently welcomed two new Board members – John Wilkins and Susan McCormack – and will continue to search for individuals who will bring the time, talent and treasure that we will require in the years ahead. As the first affected individual to sit on the Board, John brings a unique insight into the issues facing the Barth Syndrome Foundation and its primary constituents. Susan McCormack, whose younger daughter is a carrier of Barth syndrome and who brings a special focus on carriers, is a portfolio manager with a prominent investment management firm in Boston.

2010 also saw the departure of our first Executive Director, Linda Stundis. In the two years she was with us, Linda accomplished much including a rationalization of our international affiliate licensing agreements, the move of the Barth Syndrome Registry and Repository from the University of Florida to Children's Hospital in Boston, the development of our employee handbook, our initial approval by the Better Business Bureau, and the initial planning for the 2010 International Barth Syndrome Conference, among other things. We too learned much from Linda, and the search for her replacement is now underway.

Conclusion

Like those of the last decade, our progress and accomplishments over the past year have been the result of extraordinary commitment across the BSF community—our families, in their compassionate support of one another as well as their tireless fundraising efforts to support our programs and mission; our physicians and scientists who are successfully challenging the limitations of Barth syndrome science and medicine to date; and of course our donors, whose funding has leveraged the passion of our families and the dedication of our researchers, and in so doing, brought us closer to our vision of a world in which Barth syndrome no longer causes suffering or loss of life.



Stephen B. McCurdy
Chairman

In Loving Memory:

Phillip Brown (United Kingdom) — *May 22, 2005 – September 29, 2009*
Michael Bowen (United States) — *December 7, 1986 – December 9, 2009*
Jamal Thomas (United States) — *August 23, 1984 – December 23, 2009*
Jack Bradley Michael Reddin (United Kingdom) — *December 22, 2009 – December 29, 2009*
Zachary Basilo (Canada) — *April 28, 2009 – May 26, 2010*
Ben Thorpe (South Africa) — *September 2, 1994 – November 17, 2010*

BTHS Timeline — BSF ~ A Decade of Discovery

1979		Described Barth syndrome (BTHS) as X-linked recessive cardiomyopathy with abnormal mitochondria.
1981 & 1983		Fully described BTHS as an X-linked mitochondrial disease affecting cardiac muscle, skeletal muscle and neutrophil leucocytes. Barth PG, Scholte HR, Berden JA, Van dK-VMJM, Luyt-Houwen IE, Van tV-KET, Van dHJJ, Sobotka-Plojhar MA. J Neur Sc 1983; 62:327-355.
1991		Found 3-methylglutaconic aciduria to be a clinical biochemical marker for BTHS. Kelley RI, Cheatham JP, Clark BJ, Nigro MA, Powell BR, Sherwood GW, Sladky JT, Swisher WP. J Pediatr 1991; 119(5):738-747.
1995		Reported that G-CSF can be used successfully to treat neutropenia in BTHS. Cox GF, Pulsipher M, Rothenberg M, Korson M, Kelley RI. Am J Hum Genet 1995; 57(Suppl):A177.
1996		Discovered *tafazzin* (*TAZ*) gene on the distal arm of Xq28. Bione S, D´Adamo P, Maestrini E, Gedeon AK, Bolhuis PA, Toniolo D. Nat Genet 1996; 12(4):385-389.
1997		Documented heart transplantation as being successful in BTHS patient. Adwani SS, Whitehead BF, Rees PG, Morris A, Turnball DM, Elliott MJ, de Leval MR. Pediatr Cardiol 1997; 18(2):143-145.
1998		Shown that female carriers of BTHS are asymptomatic due to X-chromosome inactivation. Orstavik KH, Orstavik RE, Naumova AK, D´Adamo P, Gedeon A, Bolhuis PA, Barth PG, Toniolo D. Am J Hum Genet 1998; 63(5):1457-1463.
1999		Discovered higher-than-expected unrelated BTHS cases in one hospital in Bristol, UK, indicating an under-diagnosis. Cantlay AM, Shokrollahi K, Allen JT, Lunt PW, Newbury-Ecob RA, Steward CG. J Pediatr 1999 Sep;135(3):311-5. Erratum in : J Pediatr 2000 Jun;136(1):136.
2000		Held first BTHS Family Gathering. June 2000; Baltimore, MD. (Barth Syndrome Family Network) Sue Wilkins, Shelley Bowen, Anna Dunn
www.barthsyndrome.org		Obtained domain for Barth Syndrome Foundation, Inc.
		Barth Syndrome Foundation, Inc. officially incorporated.

(cont'd) 2000		Established BSF Board of Directors and elected Shelley Bowen, Steve McCurdy, Kate McCurdy, Sue Wilkins and Anna Dunn.
		Demonstrated that *tafazzin* is involved in cardiolipin remodeling in BTHS. Vreken P, Valianpour F, Nijtmans LG, Grivell LA, Plecko B, Wanders RJA, Barth PG. Biochem Biophys Res Comm 2000; 279(2):378-382.▼
2001		Obtained 501(c)(3) status.
		Initiated the design of BSF's logo.
		Established BSF Scientific & Medical Advisory Board.
		Presented clinical course and treatment of neutropenia in BTHS patients. Zeidler C, Barth PG, Bonilla MA, Bolyard AA, Boxer L, Cottle T, Dale DC, Donadieu J, Fier C, Freedman M, Kannourakis G, Kinsey S, Liang B, Schwinzer B, Welte K, Cham B. Blood 2001; 98(11):300a.
		Published first issue of BSF newsletter.
2002		Co-sponsored BTHS Cardiomyopathy Research Study with Drs. Byrne, Spencer, Thompson & Spevak.*▼
		Created listservs for BSF community.
		Developed relationships with ORD and NINDS (NIH).
		Launched BSF Research Grant Program.
		Received NIH funding in support of BSF 2002 Conference.
		Held 1st International Scientific, Medical & Family Conference. October 2002; Baltimore, MD.
		Examined how BTHS neutrophils function and excluded bone marrow failure and early clearance of cells as explanations for BTHS neutropenia. Kuijpers, TW. Eur J Pediatr 2002 Oct;161 Suppl 1:S75-82. Epub 2002 Sep 13.

2003

Awarded 5 research grants ($148,942).

Media coverage: BTHS/BSF featured in Readers Digest. Article titled "*Saving Michael Bowen.*"

Translated BTHS related content on website into 13 foreign languages.

Barth Syndrome Trust (UK & Europe) established.

BSF of Canada officially incorporated.

Documented phospholipid abnormalities in children with BTHS.

Schlame M, Kelley RI, Feigenbaum A, Towbin JA, Heerdt PM, Schlieble T, Wanders RJ, DiMauro S, Blanck TJ. J Am Coll Cardiol 2003 Dec 3;42(11):1994-1999. ▼

Constructed a *TAZ*1 yeast mutant model.

Gu Z, Valianpour F, Chen S, Vaz FM, Hakkaart GA, Wanders RJA, Greenberg ML. Mol Microbiol 2004 Jan; 51(1):149-158. *

2004

Awarded 5 research grants ($173,760).

Genetic Alliance

BSF highlighted as "model" member group by Genetic Alliance.

First BTHS child diagnosed *in utero*.

Joined National Health Council.

Held 2nd International Scientific, Medical & Family Conference, BTHS Clinic, and SMAB Meeting. July 2004; Lake Buena Vista, FL.

2005

Established Barth Syndrome Medical Database & BioRepository @ Univ. of FL.

BSF representative invited to participate in NHLBI Working Group - Cardiomyopathies in Children with Rare Diseases.

Awarded 4 research grants ($157,000).

Suggested that only the full-length and exon 5-deleted mRNAs of the *tafazzin* gene are important.

Gonzalez IL. Am J Med Genet A. 2005 May 1;134(4):409-14.*

Described risk of serious arrhythmias and sudden cardiac death in BTHS adolescents.

Spencer CT, Byrne BJ, Gewitz MH, Wechsler SB, Kao AC, Gerstenfeld EP, Merliss AD, Carboni MP, Bryant RM. Pediatr Cardiol. 2005 Sep-Oct;26(5):632-7. *▼

BTHS featured on Discovery Health Mystery Diagnosis. Episode titled "*Blood Brothers.*"

2006

Hired BSF Science Director, Matthew J. Toth, PhD.

Strauss et al., and Degli Esposti et al., independently created zebrafish knock-in models of BTHS; Strauss demonstrated that G4.5 gene is essential for normal cardiac development in zebrafish.

Khuchua Z, Yue Z, Batts L, Strauss AW. Circ Res. 2006 Jul 21;99(2):201-8. Epub 2006 Jun 22.*

Awarded 5 research grants ($163,801).

BBB ACCREDITED CHARITY bbb.org/charity

Established membership with BBB Wise Giving.

Documented cardiac and clinical phenotype in BTHS.

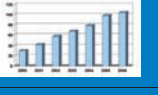
Spencer CT, Bryant RM, Day J, Gonzalez IL, Colan SD, Thompson WR, Berthy J, Redfearn SP, Byrne BJ. Pediatrics. 2006 Aug;118(2):e337-46. Epub 2006 Jul 17.*▼

Received funding from NHLBI and ORDR (NIH) in support of BSF 2006 Conference.

Held 3rd International Scientific, Medical & Family Conference, BTHS Clinic, and SMAB Meeting. July 2006; Lake Buena Vista, FL.

Created a Drosophila (fruit fly) model of BTHS.

Xu Y, Condell M, Plesken H, Edelman-Novemsky I, Ma J, Ren M, Schlame M. Proc Natl Acad Sci USA. 2006 Aug 1;103(31):11584-8. Epub 2006 Jul 19. *



Total number of living individuals diagnosed with BTHS and members of BSF surpassed 100 count.

Created Human *Tafazzin* (*TAZ*) Gene Mutation and Variation Database.

Gonzalez, IL.

Shown that BTHS-causing mutations modeled in yeast display mislocalization of the *tafazzin* protein.

Claypool SM, McCaffery JM, Koehler CM. J Cell Biol 2006 Jul 31;174(3):379-390.

Awarded 8 research grants ($320,000).

2007

Built on prior research of 2001 and more fully documented normal verbal but lower mathematical and visual spatial skills in BTHS patients.

Mazzocco MMM, Henry AE, Kelley RI. J Dev Behav Pediatr 2007 Feb;28(1):22-30. *▼

Presented testimony at Social Security Disability Hearing on Compassionate Allowances.

Held SMAB+ Meeting.

2008

Reported on development of BTHS screening using bloodspots and HPLC tandem mass spectrometry.

Kulik W, van Lenthe H, Stet FS, Houtkooper RH, Kemp H, Stone JE, Steward CG, Wanders RJ, Vaz FM. Clin Chem. 2008 Feb;54(2):371-8. *▼

Awarded 9 research grants ($333,000).

Held 4th International Scientific, Medical & Family Conference, BTHS Clinic, and SMAB Meeting. July 2006; Clearwater, FL.

Awarded first-ever Varner Award for Pioneers in Science and Medicine to Peter G. Barth, MD, PhD and Richard I. Kelley, MD, PhD.

Commissioned TaconicArtemis GmbH to create *tafazzin* "knockdown" mouse.

2009

Media coverage: BTHS/BSF featured on the TODAY Show and in PARADE Magazine.

Met all 20 Better Business Bureau Charity Standards.

Awarded 4 research grants ($143,978).

Held Genetic Diagnosis of BTHS meeting with all three US CLIA labs.

First case of BTHS female patient. Poster presented at 11th International Congress on Inborn Errors of Metabolism Meeting. Molecular Genetics and Metabolism, Volume 98, Issues 1-2, October 2009, Pages 89-118.

Held SMAB+ Meeting.

2010

Acknowledged BSF 10th anniversary ... a decade of community, education, and discovery.

Distributed *tafazzin* "knockdown" mouse to five laboratories for detailed analysis. (Four US labs and one in Germany.)

Barth Syndrome Registry and Repositiory moved to Children's Hospital, Boston, MA.

BSF representative invited to serve on planning committee of ORDR Uniting Rare Disease Research: The Intersection of Patient Registries, Biospecimen Repositories, and Clinical Data Workshop.

BSTrust received NHS funding in support of Barth Syndrome Service Clinic in Bristol, England.

Awarded 7 research grants ($267,672).

Received funding from NHLBI and ORDR (NIH) in support of BSF 2010 Conference.

Held 5th International Scientific, Medical & Family Conference, BTHS Clinic, and SMAB Meeting. July 2010; Orlando, FL.

Awarded 2010 Varner Award for Pioneers in Science & Medicine to Daniela Toniolo, PhD and posthumously to Peter Vreken, PhD.

Documented BTHS as being devastating *in utero* and under-diagnosed as cause of male fetus loss.

Steward CG, Newbury-Ecob RA, Hastings R, Smithson SF, Tsai-Goodman B, Quarrell OW, Kulik W, Wanders R, Pennock M, Williams M, Cresswell JL, Gonzalez IL, Brennan P. Prenat Diagn. 2010 Oct;30(10):970-6. *▼

BTHS clinical trial announcement made on major website (ClinicalTrials.gov).*▼

Exercise Training in Barth Syndrome. Principal Investigator: William T Cade, PT, PhD Washington University School of Medicine.

*Publications that acknowledge financial support contributed by BSF and/or BSF affiliates.

▼Publications that acknowledge biological samples (and/or information) from Barth families, the Barth Syndrome Medical Database and BioRepository (BRR), and/or BSF affiliates.

To download a PDF version of BTHS bibliography, go to: http://www.barthsyndrome.org/english/View.asp?x=1356.

Our Mission

Today, Barth syndrome (BTHS) is a rarely understood, frequently fatal, genetic disorder primarily affecting males. The Barth Syndrome Foundation is an engaged, global community whose mission is...

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

Our Values

- We will insure that BSF means: Credibility, Integrity, Professionalism, and Compassion.
- We will be respectful of the time and talents we are offered and good stewards of the resources we are given.
- We value collaboration and constantly seek to improve by listening to and learning from others.
- When representing BSF we place the interests of all those affected by Barth syndrome above the interest of any individual.
- We will never ever give up!

What is Barth syndrome (BTHS)?

Barth syndrome (BTHS; OMIM #302060) is a rare but serious genetic disorder primarily affecting males across different ethnicities. It is caused by a mutation in the *tafazzin* gene (*TAZ*, also called G4.5), resulting in a complex inborn error of metabolism.

Though not always present, cardinal characteristics of this multi-system disorder often include combinations and varying degrees of:

- Cardiomyopathy (dilated or hypertrophic, possibly with left ventricular noncompaction and/or endocardial fibroelastosis)
- Neutropenia (chronic, cyclic, or intermittent)
- Underdeveloped skeletal musculature and muscle weakness
- Growth delay (abnormal growth pattern, similar to but more severe than constitutional growth delay)
- Exercise intolerance
- Cardiolipin abnormalities
- 3-methylglutaconic aciduria

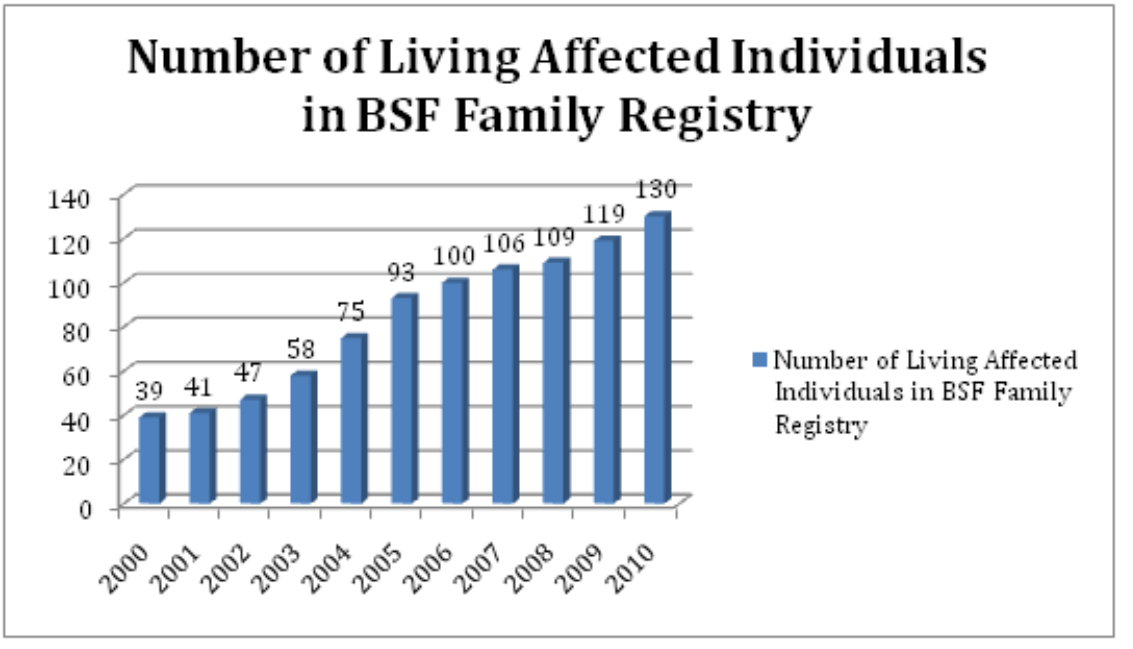
Family Services 2010 *(Valerie "Shelley" Bowen, President)*

The Barth Syndrome Foundation (BSF) Family Services program remains as relevant today as it was a decade ago when it was first established. While Barth syndrome remains a threat to human lives, it is no longer considered a uniformly fatal disease in infancy.

Our efforts have provided educational resources to transform families from powerless by-standers to empowered advocates. How do we do that? We pay attention. Our e-mail based listserv provides families with a tool to pose probing questions to our members around the world. Geographical boundaries are no longer a barrier to obtain knowledge. With one click, an inquiry goes to families, scientists and clinicians around the world. Our families regard the listserv as a "lifeline" to gain information.

(Cont'd on page 8)

(Cont'd from page 7)



The listserv is regarded as a "pipeline" of information about the disease among expert clinicians and scientists in our community. We carefully track the information being shared. In so doing, we are able to identify potential trends of the disease. We appreciate information exchange as non-stagnant data. When potential trends are brought to the fore, we then utilize a state of the art survey software platform to obtain further data. This information is then filtered to the doctors and scientists widely considered by their peers as experts in the various aspects of Barth syndrome for feedback.

Information exchange among family members have propelled investigations into the issues at hand and have ultimately made a difference in propelling the mission of our organization and further validating our belief that lives can be saved through a diagnosis and informed decision-making.

The listserv is supplemented by the personal support provided by Shelley Bowen and the Family Services team who are available to any affected family for ongoing issues and at times of medical crisis through a simple phone call to BSF. The BSF phone line rings virtually every day throughout the year and responses, if not immediate, almost always take place within 24 hours. While careful not to provide medical advice, Family Services helps families through the complex process of being effective advocates with their child's multiple medical specialists. Often, the help comes in the form of a simple connection formed between the child's physicians and the world's experts in Barth syndrome. In every case, however, worried families are comforted by having a warm, caring and experienced person to talk to and to help make sense of a stressful and confusing situation.

(Photos courtesy of Amanda Clark ~ BSF 2010 Conference, July 2010, Orlando, FL.)

Barth Syndrome Posters Presented at Scientific/Medical Conferences in 2010

A Novel Missense Mutation M185V in the *TAZ* Gene Associated with Atypical Barth Syndrome
Fan Y, Chang R, Fox M, Westerfield BA, Steller J, Batra AS, Wang RY, Dipple K, Gallant N, Pena LS, Wang H, McCabe ER, Kimonis VE
The American Society of Human Genetics 60th Annual Meeting, Washington. DC, November 2-6, 2010.

Characterization of an shRNA-Mediated *Tafazzin* Knockdown Mouse Model for Barth Syndrome
Soustek MS, Falk DJ, Lewin AS, Byrne BJ
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Role of Cardiolipin and Phosphatidylethanolamine in Mitochondrial Fusion
Joshi AS, Greenberg ML
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Investigating Barth Syndrome: The Role of Cardiolipin in Maintenance of Mitochondrial Morphology
Fei N
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

A Reversed-phase LC-MS/MS Cardiolipin Method for the Diagnosis of Barth Syndrome Suitable for Use in a Routine Metabolic Lab
Bowron A, Frost R, Heales S, Steward CG
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Genetic and Viral Genome Analysis of Childhood Cardiomyopathy: The PCMR/PCSR Experience
Towbin JA, Sleeper L, Jefferies JL, Colan S, Webber SA, Canter CE, Hsu DT, Ware SM, Wilkinson JD, Orav EJ, Lipshultz SE
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Lipidomic and Bioenergetic Effect of Cardiac-specific Overexpression of Cardiolipin Synthase
Kiebish MA, Yang K, Sims HF, Mancuso DJ, Zhou Z, Guan S, Gross RW, Han X
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Impact of Sample Collection on *Tafazzin* mRNA Variants
Kirwin SM, Swain SE, Manolakos AL, Gonzalez IL
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Fetal Barth Syndrome: A Case of Progressive Cardiomyopathy in Utero
Tsai-Goodman B, Martin RP, Steward CG
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Full-Length Human *Tafazzin* Protects the Human Cells Fom Serum-Withdrawn Apoptosis
Xu Y, Schlame M
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Loss of Mitochondrial Anionic Phospholipids Leads to Perturbation of Mitochondrial/Cellular Iron Homeostasis
Patil VA, Gohil VM, Gupta S, Li G, Greenberg ML
2010 Barth Syndrome International Conference, Orlando, FL, July 26-31, 2010

Barth Syndrome in a Female Patient
Cosson L, Toutain A, Simard G, Paoli F, Kulik F, Vaz FM, Blasco H, Chantepie A, Labarthe F
2010 Barth Syndrome International Conference, Orlando, FL, Jul 26-31, 2010

Analysis of *TAZ* (*tafazzin*) and LDB3 (LIM domain-binding3/Cypher/ZASP) Genes in Left Ventricular Noncompaction
Villard E, Habib G, Donal E, Eicher JC, Pascal C, Isnard R, Dilanian G, Komajda M, Charron P. Archives of Cardiovascular Diseases Supplements 2010.

Genetic and Viral Genome Analysis of Childhood Cardiomyopathy: The PCMR/PCSR Experience
Towbin JA, Sleeper L, Jefferies JL, Colan S, Webber SA, Canter CE, Hsu DT, Ware SM, Wilkinson JD, Orav EJ, Lipshultz SE
American College of Cardiology 59th Annual Scientific Session, Atlanta, GA, March 15, 2010.

International Scientific, Medical & Family Conference 2010

In 2010, the 5th biennial Barth Syndrome International Scientific, Medical & Family Conference was held in Orlando, Florida, on July 26—31. The Scientific and Medical Sessions of the Conference were 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. Nearly 350 Barth syndrome families, physicians, scientists, and volunteers from around the globe were in attendance. There is no BSF program that has a greater impact on our mission. Scientists were inspired not only by cutting-edge science presented by researchers from around the globe, but also by witnessing the remarkable spirit of our families and by meeting the boys who benefit from their research. Physicians were able to see and hear about a number of affected individuals, thus helping improve treatment approaches to the disorder. Families were empowered as advocates, as their perspectives were not only heard but taken to heart. Relationships were forged and research collaborations were born. Ideas flourished. Commitment to the cause deepened. An extraordinary sense of community was strengthened.

Barth Syndrome Clinics

Also unique to this conference series is the information-gathering session or "clinic" that is provided by Barth syndrome (BTHS) physicians, healthcare workers and researchers. This "clinic" serves at least two purposes: (1) it allows the efficient collection of physiological data and historical medical information from patients with this rare disease; and (2) it provides opportunities for patients and patient family members to meet with physicians who have substantial experience in treating BTHS individuals. In 2010, many of the BTHS individuals who attended the "clinic" participated in six distinct IRB-approved protocols. Most of the data collected are expected to lead to publications and/or ultimately to be available through the Barth Syndrome Registry and Repository (BRR) which is open to all interested researchers.

Photos of Barth Syndrome Clinics (Cardiology and Neurology) at BSF's 2010 Conference, July 2010, Orlando, FL. (Photos courtesy of Cherie Schrader.)

Family Sessions

The Family Sessions included presentations of the most up-to-date information on Barth syndrome (BTHS). There were updates on research, cardiac and arrhythmia concerns, hematology and clinical manifestations of the syndrome and how to treat them. Families had the opportunity to ask questions to gain a better understanding of living with the complex symptoms of BTHS and to learn coping mechanisms and strategies from those who know best—the families themselves. There also were programs specifically designed for and by the young attendees. New participants met others and formed lasting relationships, and experienced attendees had ample time to strengthen friendships and form new bonds.

Photo of Family Session at BSF's 2010 Conference, July 2010, Orlando, FL. (Photo courtesy of Cherie Schrader.)

There were nearly 50 affected families in attendance, 12 of whom were first-time conference attendees. Many had travelled great distances to meet with world-renowned experts in the field of BTHS to gain the most up-to-date information about treatments and research into this disorder.

Scientific and Medical Sessions

This was the fifth international conference hosted by the Barth Syndrome Foundation (BSF). Conferences are held every two years, and 2010 marked the 10th anniversary of the founding of BSF by family members of Barth syndrome (BTHS) individuals. Many of the speakers over the two days of the Scientific/Medical (Sci/Med) sessions of the Conference were BSF research grant recipients. The chairpersons for the Sci/Med sessions were: Richard I. Kelley, MD, PhD (Johns Hopkins University, Baltimore, MD); Michael Schlame, MD (New York University, New York, NY); Barry J. Byrne, MD, PhD (University of Florida, Gainesville, FL); and Miriam Greenberg, PhD (Wayne State University, Detroit, MI).

(L-R) Photos of Sci/Med Session and Poster Session at BSF's 2010 Conference, July 2010, Orlando, FL. (Photos courtesy of Cherie Schrader.)

Also included this year was a keynote lecture, "The Pathophysiology of Mitochondrial Disease," which was delivered by Professor Douglas C. Wallace, PhD, Director of the Center of Mitochondrial and Epigenomic Medicine, Children's Hospital of Philadelphia and University of Pennsylvania. The Sci/Med sessions of the 2010 Conference were 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.

Animal Models of Barth Syndrome

Animal models of a human disease are often valuable tools in finding and developing clinical treatments, and leading off the Science/Medicine sessions were the first reports of the mouse model of Barth syndrome (BTHS) developed by BSF. These initial reports of the Barth syndrome mouse were quite encouraging. Zaza Khuchua, PhD (Cincinnati Children's Hospital Medical Center, Cincinnati, OH), and Michael A. Kiebish, PhD (Washington University School of Medicine, St. Louis, MO) revealed that this "knockdown" mouse model possesses the expected biochemical abnormalities and also eventually develops the heart problems characteristic of BTHS.

Other animal models, including fruit flies (Mindong Ren, PhD, New York University, New York, NY) and rat models of heart failure (Genevieve Sparagna, PhD, University of Colorado, Boulder, CO), continue to add to our understanding of BTHS and provide new ways to think about potential treatments. BSF intends to distribute these BTHS mice to any investigator, and we expect a substantial increase in the number of new BTHS researchers since they will now have a powerful model system in which to test their hypotheses. Currently these animals are being studied in five laboratories in the US and Europe.

Barth Syndrome Pathophysiology

Barth syndrome (BTHS) is considered a unique mitochondrial disease. Mitochondria are those distinct parts of the cell which provide it with energy; defective mitochondria have been implicated as the cause of several human diseases and as one of the main causes of the natural aging process. Using exercise as therapeutic treatment, Mark Tarnopolsky, MD, PhD (McMaster University, Hamilton, Ontario) showed that mitochondrial DNA deletions in elderly people can be reversed by exercise. Dr. Tarnopolsky then discussed what this may imply for BTHS. In one of the first clinical research projects funded in part by the BSF Research Grant Program, Todd Cade, PT, PhD (Washington University School of Medicine, St. Louis, MO) is now testing whether supervised aerobic exercise training (cardiac rehabilitation) is beneficial for BTHS individuals.

Dr. Cade, along with Carolyn Spencer, MD and Amy Roberts, MD (both at Children's Hospital Boston, Boston, MA) also presented the unique physiological characteristics of BTHS individuals. These physiological data, much of which have been collected at the conference "clinic," provide us with a better understanding of this disorder. The Barth Syndrome Registry and Repository (BRR), which is supported by BSF and now also by the Children's Hospital Boston, will collect and store these data, other relevant medical information and numerous biological samples for researchers to use.

(Cont'd on page 12)

(Cont'd from page 11)

Of particular note for the BSF community was a presentation by Colin Steward, FRCP, FRCPCH, PhD (Bristol Royal Hospital for Children, Bristol, England). Dr. Steward has found many previously unrecognized cases of BTHS in the Bristol area of England by pursuing the neutropenia aspect of this disease and by doggedly investigating unexplained male fetal deaths in several affected families (Barth syndrome: an X-linked cause of fetal cardiomyopathy and stillbirth. See page 6 for complete journal citation.) Dr. Steward, along with the assistance of Michaela Damin and the Barth Syndrome Trust (the UK affiliate of BSF), has set up a National Specialized Service for Barth syndrome within the National Health Service in the UK, and he provided insights for establishing a similar group in the US.

Mitochondrial Dysfunction and Its Impact on Human Disease
Lipids, *Tafazzin*, and Mitochondrial Metabolism in Barth Syndrome

Human diseases that resemble BTHS also have a lot to teach us. Charles Hoppel, MD (Case Western Reserve University, Cleveland, OH) provided an overview of mitochondrial diseases by focusing on oxidative phosphorylation defects. Robert E. Jensen, PhD (Johns Hopkins University, Baltimore, MD) illuminated the important parallels between Barth syndrome (BTHS) and Dilated Cardiomyopathy with Ataxia (DCMA) and discussed how mitochondrial protein transport may link the common symptoms of these two genetically distinct but similar, rare conditions. By relating what we know about other human diseases that share characteristics with BTHS, we may be able to formulate treatment ideas for both BTHS and for these other diseases.

The number and breadth of the Science/Medicine presentations show that BTHS research has come of age. These sessions featured presentations ranging from lipid biochemistry; the composition, intermediary metabolism, and dynamic movements of mitochondria; and the screening for genes/compounds that reverse the genetic defects of the BTHS mutation. Most of these presentations were made possible, in part, by funding provided by BSF and our affiliates through its Research Grant Program. The eight years of this grant program, along with the conference "clinic," the BRR, and the dedicated efforts/publications of BTHS researchers and BSF members have provided the scientific/clinical foundation to guide investigators to make the progress that will translate into real clinical benefits.

In addition to the Science/Medicine presentations, 12 posters were presented in a separate session that was well received by both the scientific/medical attendees and by the families of BTHS individuals. The interactions between these two groups are extremely important as both groups get to know and appreciate the details and the problems each face—a perspective that often is lacking in other science/medicine-oriented meetings.

Varner Award for Pioneers in Science and Medicine

The Varner Award for Pioneers in Science and Medicine was awarded to Daniela Toniolo, PhD, and posthumously to Peter Vreken, PhD. Dr. Toniolo (Research Director, National Research Council of Italy, DIBIT-San Raffaele Research Institute, Milan, Italy) was recognized for her discovery of the *tafazzin* gene in 1996, and the late Dr. Vreken (Academic Medical Center, University of Amsterdam, Amsterdam, The Netherlands) was recognized as the first to publish on the cardiolipin abnormalities of BTHS individuals in 2000. It was particularly appropriate to mark these significant scientific milestones at BSF's 10th anniversary celebration.

John Wilkins and Jessica Wiederspan (Wilkins) presented the 2010 Varner Award for Pioneers in Science and Medicine to Dr. Daniela Toniolo. (Photo courtesy of Cherie Schrader.)

In Summary

The 2010 Conference included the greatest number of speakers in its history, only some of whom could be mentioned here due to space considerations. The Scientific and Medical (Sci/Med) sessions were packed with new information and new developments. All of the individual presentations, for both the Sci/Med sessions and the Family sessions, were recorded on DVDs and are available for a nominal cost by contacting BSF (www.barthsyndrome.org). Given the breadth and quality of the work presented at this latest Conference, the next meeting in 2012 is sure to reveal even further progress towards a specific treatment for this complex rare disease.

Conference Attendee Testimonials

"Having never attended a Barth syndrome conference before, I could never have anticipated the sense of community and urgency that encompasses the Barth Syndrome Foundation's families and dedicated scientists. In my opinion, no conference exists that integrates and recapitulates the emotion and purpose of synergistically merging the efforts of the research community with that of the passionate goal of the families toward developing a therapeutic strategy to treat young boys/men affected with this disease. You might walk into the conference as a scientist, but as I found, you walk out of the conference as an adopted family member working for the same cause and battling for the same goal. ..." *~ Michael Kiebish, PhD, Xianlin Han Laboratory, Washington University School of Medicine, St. Louis, MO*

"A huge thank you to all the volunteers who made this conference a spectacular event. I wish I could explain to all ya'll (LOL) how much it meant to our family, how much it touched us, how much it taught us, and how grateful we are having met everyone. When our son's doctors first told us that our son had a rare disorder called Barth syndrome, I was very confused, angry and frightened. I reached out to this community and was immediately welcomed with open arms. I was given wonderful advice and many promises that in this community WE DO NOT GIVE UP on OUR BOYS! How true I have found this to be." *~ Brie, BTHS Family, Kentucky, USA*

"It was my first time at an International Meeting of the Barth Syndrome Foundation, 14 years after identification of the gene, and it was a very exciting experience. I had followed the work done by the Foundation through the BS Journal that I received regularly, but the active participation in the meeting of so many scientists and of many patients and family members was very impressive. I think the scientific results presented at the meeting and the general atmosphere were unique, and they represented the best reward for our work. Thanks again for inviting me and giving me the opportunity to see how sometimes the results of a scientific achievement can go beyond a good publication." *~ Daniela Toniolo, PhD, Division of Genetics & Cell Biology, San Raffaelel Scientific Institute, Milan, Italy*

"I am writing this on our drive home, with the bittersweet feeling of the extraordinary week we spent together laughing, crying, supporting and inspiring each other. I am going home with a "high" unmatched by any other experience I've had. There is so much that we need to process and reflect upon from this past week; from the informative lectures, the private conversations and life-long bonds that strengthen with each conference. We want to genuinely thank ALL who made this week a memorable one. It may be hard to envision 2012 now, but I genuinely can't wait to reunite again with our Barth family." *~ Amer, BTHS Family, Florida, USA*

"I was very pleased to attend the 5th BSF meeting to talk about my research on X-chromosome inactivation in female carriers of Barth syndrome. I have been a clinical geneticist for almost 40 years but previously only met one family with boys with Barth syndrome. Therefore, I greatly enjoyed meeting all the boys and their families and hearing their stories, which they presented so bravely to the audience. I was also very impressed by the scientific quality of the meeting. The conference was an outstanding example of how efforts initiated by parents can contribute to increased knowledge about this rare and puzzling disorder." *~ Karen Helene Orstavik, Department of Medical Genetics, Oslo University Hospital, Oslo, Norway*

"Our family made the trip from Scotland to Orlando to attend the 2010 conference because it is the one place where other people "get" what Barth syndrome is and what it means to your son and family. You can completely and utterly immerse your full family in this wonderful group of people. We always find that we can learn so much from the other families that attend, and the time spent at the clinics and family sessions are just as important as time spent around the pool with the other families sharing experiences, love, information and supporting one and other. If your family has not attended one of these conferences then I would urge you to try to, as you cannot buy the fountain of information that all the families are willing to share with you and your family. It truly is a remarkable group to be part of, and my family is very grateful to each and every person who works so tirelessly for the benefit of us all." *~ Allanna, BTHS Family, Scotland*

"I have been involved with the Barth boys now for over six years, and I am still learning. There are so many questions, and the answers are coming through but slowly. I really enjoyed the Barth Syndrome Foundation Conference in Florida, as it gave me the opportunity to meet other people who are also interested in helping these boys, from scientists to medical doctors to parents, friends and family! I have never come across any medical meeting where scientists, medical doctors and patients sit next to each other and learn from each other. A real gem of an opportunity!" *~ Beverly Tsai-Goodman, BM, MD, FRCP, PG Cert Med Ed, Department of Paediatric Cardiology, Bristol Royal Hospital for Children, Bristol, England*

"I would sincerely like to thank BSF for inviting me to speak at the 2010 meeting in Orlando. This was my first BSF meeting and it was a very enlightening academic and scientific experience since I had very little exposure to Barth syndrome and the underlying pathology prior to attending this conference. More importantly, although I have attended numerous scientific meetings throughout my career, I have never had the opportunity to interact with family members and individuals with a particular disease affliction. This interaction had a profound impact on me, and certainly put all of the basic science that we do in the laboratory into perspective. I hope to have the opportunity to be involved with this conference and BSF in years to come! *~ Peter Adhihetty, PhD, Department of Applied Physiology and Kinesiology, University of Florida, Gainesville, Florida, USA*

Science and Medicine 2010 *(Matthew J. Toth, PhD, Science Director)*

Research Grant Program

The Barth Syndrome Foundation (BSF) awarded seven new research grants for over $277,968 in early 2010 (2009 grant cycle) and another seven grants totaling $273,770 in early 2011 (2010 grant cycle). The following is a summary of the main focus of each grant awarded in the 2009 cycle. The breadth of these awards clearly demonstrates the progress being made and increased exposure of Barth syndrome (BTHS) research within the scientific and medical communities. These 2009 grant recipients are testing ideas or measuring parameters that will lead to a better understanding and perhaps a better treatment of BTHS individuals. All of the recipients are building on previous work performed in their laboratory or at the clinics of the BSF biennial conferences.

With the completion of the 9th cycle of BSF's Research Grant Program, it is a good time to reflect on its value towards accomplishing the goals of our unique organization. Before this grant program began, knowledge about Barth syndrome, its causes, or its treatment was scarce or nonexistent. The original publications by Dr. Peter Barth in the early 1980s were supplemented occasionally over the next two decades by several reports which included the identification of the defective gene—*tafazzin*. Since those early days, over 160 publications have appeared in the scientific/medical literature (125 since 2002) of which 55 acknowledge the financial support of the BSF and its affiliates. Without the support of the BSF Research Grant Program which began in 2002, how many fewer manuscripts would have been published? How many researchers would even know about this unique mitochondrial disease with its complex interplay of an enlarged and weakened heart, low white blood cell count, growth delay, lipid abnormalities?

(Note that throughout this article, specific BSF grant awards are shown in parentheses with the PI's name and relevant grant cycle.)

Including the 2010 cycle, the BSF has committed a total of US $ 2.0 million to its Research Grant Program—its largest yearly budget expenditure. The basic understanding of the complexities and pathologies of Barth syndrome (BTHS) is now being systematically explored by many researchers worldwide. Perhaps the easiest research projects to show impact are those involving animal models of BTHS because these genetic models are very useful in understanding the disease process and in testing new treatments. Starting with the first grant cycle, we can trace the initial development of animal models *(Strauss 2002, 2004)* which eventually culminated in a zebra fish model. A particularly valuable fruit fly model *(Ren 2004, 2006; Malhotra 2008; Xu 2009)* was then developed that shows what other genes interact with the defective *tafazzin* gene. Most recently, the mouse model was developed independently by the BSF and is being distributed to many investigators worldwide *(Khuchua 2009; Kiebish 2009; Byrne 2010; Phoon 2010; Chicco 2010)*. This mouse knockdown model is still being investigated but has already shown many interesting parallels to human BTHS. With the availability of the mouse model of BTHS, many laboratories not previously involved with BTHS research are now in a position to make contributions to our knowledge base. All of these animal models answer different questions about what BTHS is, and more importantly, how this disease may be altered to make it less deadly to BTHS individuals. In addition, rat disease models *(Sparagna 2006, 2008; Moreno-Quinn 2007)* that have similarities to BTHS have provided insights about how simple diet additions of certain oils may impact the symptoms of BTHS.

While not an animal model, the yeast model of BTHS *(Greenberg 2002, 2005, 2006, 2007, 2008, 2009, 2010; Vaz 2002; McMaster 2007)* is a workhorse for understanding many of the details of what biochemical processes are disrupted by this genetic disease. The yeast system also offers the promise of screening thousands of chemical compounds (just like what pharmaceutical companies do) so as to find compounds that may alter the disease process *(McMaster 2010)*.

In the same category of cellular models of disease like the yeast system are those mammalian cell lines that mimic the basic biochemical defect of BTHS—the defective *tafazzin* gene. While the *tafazzin* gene produces a protein that affects a fatty substance or lipid called cardiolipin, the full functional consequences of having a damaged *tafazzin* gene are still being worked out. Researchers have focused on understanding what cardiolipin is important for *(Haines 2005; Kobayashi 2005, 2006; Vaz 2006; Epand 2007; He 2007; Pu 2009)* and how defects in *tafazzin*/cardiolipin affect the health/functioning of the cell *(Hatch 2002, 2005, 2009; De Kroon 2010; van Raam 2010)*. BTHS is distinctive because it was the first human disease to be characterized as having a problem with the lipid cardiolipin. Defects in cardiolipin, however, have recently been observed in common diseases such as diabetes and heart failure making BTHS research particularly relevant to these other diseases.

Projects actually involving BTHS individuals are exceedingly hard to perform, given the complexities and costs associated with this sort of work, but these experiments directly tell us useful clinical information. Nevertheless, several researchers have made great progress in working with Barth syndrome individuals and have contributed to our clinical picture of this disease *(Spencer 2004, 2006; Storch 2005; Mazzocco 2006; Cade 2008, 2009)*. These clinical projects have described some important clinical aspects of the disorder and have shown that the metabolism of BTHS individuals is unique, which could suggest better ideas for a specific treatment. In addition, investigators of the low white blood cell count of BTHS have had an especially difficult set of obstacles to overcome in this extremely important area *(Dale 2003, 2007; Esposti 2003; Kuijpers 2003, 2007)*.

(Cont'd on page 15)

Because Barth syndrome (BTHS) is a genetic disease, the recording of genetic data so as to provide genomic insights has been supported by the BSF Grant Program *(Gonzalez 2002; Ma 2003; Taylor 2007)*. BTHS affects all races. Understanding how one's genes affect one's symptoms is important because a better understanding of the interplay of genes may hold a key to finding why some individuals do better medically than others. In addition, it is extremely important for an individual's clinical care to receive a correct diagnosis as early as possible. To address this point investigators have gone beyond the standard genetic tests and developed novel ways to better measure *tafazzin* gene expression *(Kirwin 2007)* and to measure cardiolipin *(Xu 2003; Kulik, 2005, 2006)* in blood or tissue samples taken directly from individuals. Achieving a correct diagnosis of BTHS has always been a serious problem within the undiagnosed community.

While the activities supported over the last nine years are impressive (54 grants), and the large number of researchers supported by this Research Grant Program (34 researchers) has firmly established the groundwork for making more advances in this rare disease, we need to do more. Boys and young men are still dying from BTHS, even with the best medical care! Scientifically we know far more than even a few years ago—medically we need to make more of an impact. We have the tools needed to translate what we know into new ideas for treatment.

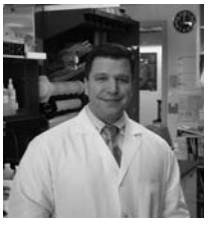
The BSF Research Grant Program started out with very few researchers even able to suggest ways to make progress on Barth syndrome research. In this last grant cycle, we had to turn down applications of merit because funds were not available. We need more good researchers. We need more support within our organization to fund applications and to implement clinical projects. We need the professional scientific/medical community to respond by funding those worthy but expensive clinical/scientific experiments that BSF-supported researchers are now applying for from the NIH and large international funding agencies.

The BSF has always committed everything it can towards advancing a treatment for Barth syndrome (BTHS). The rarity of BTHS continues to be a major obstacle to attracting researchers, though we work constantly to overcome this. In the beginning, there were few researchers. Now, success has cultivated many more researchers, each with his or her own good ideas waiting to be put to the test. The goals of the BSF will be realized by continuing with what has met with success in the past—the BSF Research Grant Program. As one of our young men with BTHS said to the researchers attending the 2010 BSF Conference, "Please give us treatments to try, and if they don't work, we'll try again. . . . Please don't let us lose any more of our family members!" We need to keep on trying.

2009 Research Grant Cycle

Zaza Khuchua, PhD, Research Associate Professor, Children's Hospital Medical Center, Cincinnati, OH
"The shRNA-mediated *tafazzin* knockdown mouse model for Barth syndrome"
Award — US $39,998 over 2-year period

Dr. Khuchua proposes to continue his analysis of the characteristics (phenotype) of the *tafazzin* knockdown mouse, the mouse line that BSF contracted with TaconicArtemis of Cologne, Germany to produce. The goal is to determine if this mouse line resembles individuals with Barth syndrome in regard to symptoms like cardiomyopathy (exercise intolerance, neutropenia, growth delay) and in regard to biochemistry like abnormal cardiolipin levels (organic aciduria, abnormal mitochondria, abnormal mitochondrial respiration, increased ROS production—complexes I, III, and V activities, supercomplex assembly, ANT activity). Dr. Khuchua has communicated preliminary results that show abnormal cardiolipin levels in at least four organs of the *tafazzin* knockdown mouse.



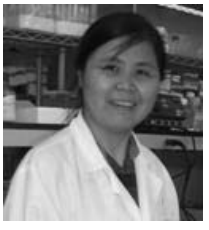
Michael A. Kiebish, PhD, Postdoctoral Research Associate, Washington University School of Medicine, St. Louis, MO
"Does cardiolipin synthase upregulation alleviate cardiolipin abnormalities and bioenergetic dysfunction in Barth syndrome?"
Award — US $40,000 over 2-year period

Dr. Kiebish proposes to analyze the characteristics (phenotype) of the *tafazzin* knockdown mouse line (supplied by BSF) and to concentrate on the changes in its lipid characteristics specifically using the "shotgun lipidomics" developed in his lab (sensitive analytical method that measures small quantities of different cardiolipin species). He intends to analyze the enzyme activities of the electron transport chain (complexes I through V) of these mice as well as to monitor the changes in heart cardiolipin. In addition, Dr. Kiebish intends to crossbreed the *tafazzin* knockdown mouse with a transgenic mouse line that overexpresses the human cardiolipin synthase gene (hCLS) and monitor for any changes in lipids or if the hCLS gene can reverse (suppress) the changes that the *tafazzin* gene knockdown causes. Interestingly, the hCLS transgenic line appears to have an increased heart fractional shortening.

(Cont'd on page 16)

Miriam Greenberg, PhD, Professor and Associate Dean, Wayne State University, Detroit, MI
"Perturbation of mitophagy in cardiolipin mutants"
Award — US $40,000 over 1-year period
**Funding for this award was provided by the Barth Syndrome Trust (UK & Europe)*

Dr. Greenberg has uncovered an interesting observation in at least two yeast strains that are mutated in genes that control cardiolipin synthesis—the pgs1 and cdrs1 genes. She found that when these genes are mutated the yeast cell displays an abnormal vacuole (enlarged) which is a subcellular organelle equivalent to the mammalian subcellular organelle called the lysosome. The lysosome is thought to be the "garbage disposal system" of the cell—the place where other subcellular organelles are disassembled and recycled for further use by the cell. These same cardiolipin mutants can be restored (suppressed) to a normal condition (wild-type) when certain genes involved in the turnover or recycling of mitochondria (mitophagy genes) are mutated or inactivated. Dr. Greenberg will determine if the *tafazzin* gene has similar properties to these other two genes.



Yang Xu, PhD, MD, Instructor, New York University School of Medicine, New York, NY
"Expression levels, localization, and function of *tafazzin* isoforms"
Award — US $38,370 over 1-year period

Dr. Xu proposes to use the newly developed antibodies (in collaboration with Dr. Steven Claypool) made against human *tafazzin* protein to gain insight into the nature of *tafazzin* isoforms of mammalian tissues. Currently we have no tool to visualize the *tafazzin* protein in human cells, and simple observations of the native (endogenous) *tafazzin* protein (Western blots) could reveal important information or suggest new hypotheses. For example, we know that human cells produce a *tafazzin* mRNA that is shorter than the predicted full length (exon 5 deletion)—does this shortened *tafazzin* mRNA (isoform) correspond to a smaller *tafazzin* protein? Dr. Xu intends to identify the different forms of the *tafazzin* protein (isoforms) that are present in different human cell lines and human tissues (including BTHS), to determine the intramitochondrial location of these isoforms, and to determine the functional differences (enzymatic) if any, between these isoforms. Dr. Xu has already provided data that shows the value of these antibodies by detecting endogenous *tafazzin* protein in human cell lines, and the absence of this protein in several Barth syndrome cell lines.

W. Todd Cade, PT, PhD, Assistant Professor, Washington University School of Medicine, St. Louis, MO
"Safety and efficacy of aerobic exercise training in Barth syndrome: a pilot study"
Award — US $39,600 over 2-year period

This clinical project is designed to determine the value of 12 weeks of supervised aerobic exercise training (cardiovascular rehabilitation) in Barth syndrome individuals. The plan is to monitor the participant's exercise performance before (most conveniently done at the BSF Conference in July, 2010) and after the supervised exercise program. From past BSF conferences, Dr. Cade and colleagues have measured several physiological indices of Barth syndrome individuals: their exercise limitations, fatigue levels, whole body oxygen consumption, cardiac performance, and skeletal muscle tissue extraction/utilization. We know that older patients with various cardiac problems show a real benefit with this type of exercise training. The hypothesis here is to find out if the same types of benefits seen in other cardiac patients are transferable to Barth syndrome individuals. Should this project demonstrate a real benefit with aerobic training, it may form the basis of a clinical recommendation for treatment.

Grant Hatch, PhD, Professor, University of Manitoba, Winnipeg, Manitoba, Canada
"Role of human monolysocardiolipin acyltransferase in Barth syndrome"
Award — US $40,000 over 1-year period
**Funding for this award was provided by the Barth Syndrome Foundation of Canada*

Dr. Hatch proposes to extend his recent publication of the newly discovered human enzyme MLCL AT1 (monolyso-cardiolipin acyltransferase) to determine if it can substitute for *tafazzin* function in several Barth syndrome cell lines (lymphoblasts). Specifically, several cell lines from Barth syndrome individuals who differ in severity of their disease will be transformed with the human MLCL AT1 mini-gene. He will determine if cardiolipin levels and mitochondrial energy production have returned to normal (wild-type) levels. He will also determine if the endogenous levels of MLCL AT1 in these same cell lines can be correlated with the disease severity. Preliminary experiments have shown that this mini-gene does elevate the cardiolipin levels in one Barth syndrome lymphoblast line and may restore some of the mitochondrial function. This avenue of investigation could serve as an alternate therapeutic approach. If we can find a way to increase MLCL AT1 expression in *tafazzin* deficient cells, it may ameliorate some of the symptoms of Barth syndrome.

(Cont'd on page 17)

William Pu, MD, Associate Professor, Children's Hospital of Boston, Boston, MA
"Analysis of metabolic abnormalities in *TAZ*-deficient cardiomyocytes"
Award — US $40,000 over 1-year period

Dr. Pu is proposing to develop two mammalian cellular models of *tafazzin*-deficient cardiomyocytes to study the effects on the citric acid cycle—the abnormality that Dr. Richard Kelley has proposed to be at the center of the pathophysiology of Barth syndrome. In the first model he will make a shRNA-knockdown construct (adenovirus vector) in rat ventricular cardiomyocytes. In the second model he will use the relatively new technology of induced pluripotent stem cells (iPSC) by taking fibroblasts from skin biopsies of Barth syndrome individuals and deriving cardiomyocytes using a series of genetic transformations. These cellular model systems will be analyzed for their citric acid cycle function and other mitochondrial properties using gas chromatography-mass spectrometry (GC-MS) and various radiolabelled tracer compounds. The expectation is that these experiments will validate the idea that abnormalities in the citric acid cycle are connected with Barth syndrome, it may lead to new ways to find therapeutic compounds, and it could provide a method to monitor treatment.

Publications about Barth Syndrome

One measure to demonstrate the progress in science and medicine is the number of peer-reviewed publications. One can show the impact of BSF and its Research Program by counting the increasing number of publications about Barth syndrome over the years and the number of those publications for which the BSF has provided grant support or contributed important information (see Figure 1). As shown by the fraction of the bar that is colored in light blue, the BSF is acknowledged in over half of the current Barth syndrome literature. Because of the rarity of Barth syndrome, often the only contact physicians or independent researchers will have is mediated through these publications. It is vital to our organization's mission that we increase these numbers so we can reach our ultimate goal. Through better awareness, better education, and better understanding of what Barth syndrome does to the individual and to their families, we will see advancements.

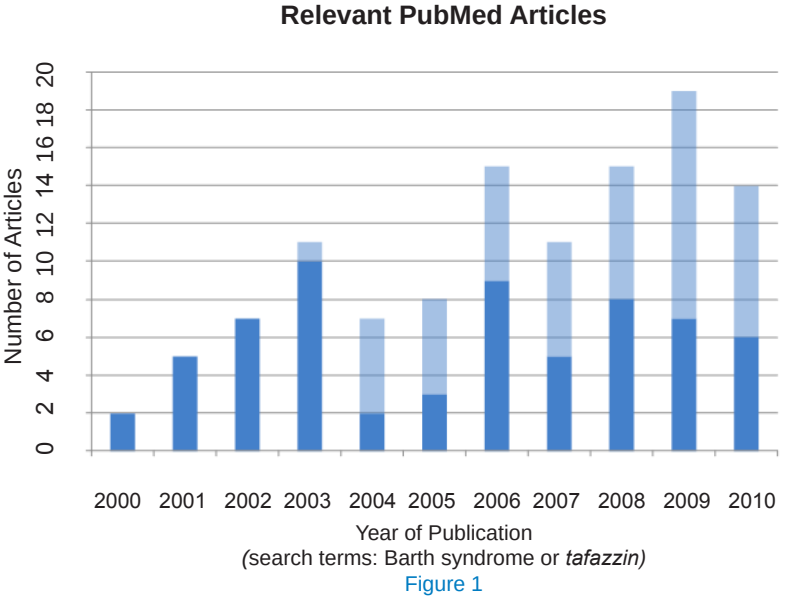


Figure 1

Government Funding of Barth Syndrome Research

One of the major goals of the BSF Research Grant Program is to provide support for investigators to gather data that will lead to funding through traditional government funding agencies like the NIH. The NIH is the largest and most valuable scientific/medical funding agency in the world and in human history. Almost every important scientific or medical advancement over the last 40 years can trace its development to the financial support of the NIH through its biomedical research program—by directly supporting research, researchers, and research institutions. Over the years the NIH, though its sub agencies like the Office of Rare Disease Research and the National Heart, Lung, and Blood Institute, has been the major provider of funds to investigators interested in Barth syndrome. The NIH funding program is vital towards obtaining the goals of BSF. Individual awards for investigator-initiated projects (often referred to as R01 grants) can reach monetary levels 8-fold of what BSF provides through its Research Program. As you can see, obtaining these important research grants is vital to our progress as well as to the professional advancement of the individual investigators.

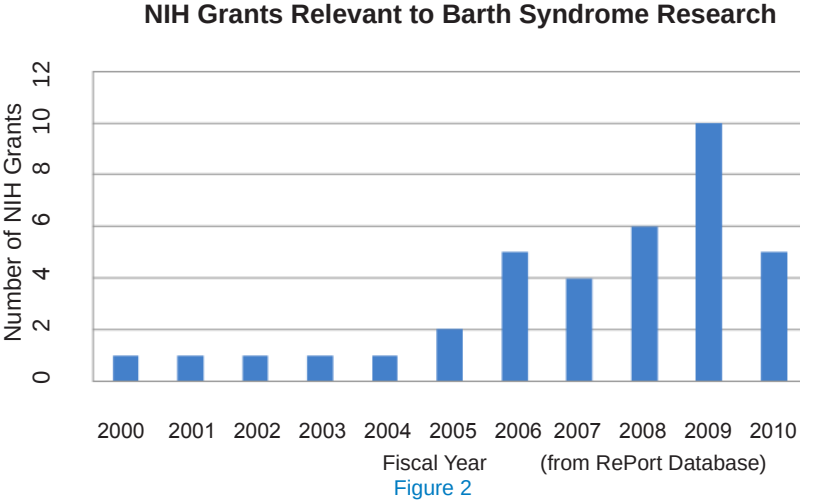


Figure 2

Over the years, the number of NIH grants for Barth syndrome research languished until 2006 when there was a significant uptick in the number of awards (see Figure 2). Since that time, there has been a sustained improvement in the number Barth syndrome-related NIH grants especially for fiscal year 2009. We like to believe that the BSF, with its Research Grant Program and by the participation of BSF individuals, is responsible for this improvement over these later years. The latest success in the number of NIH grants awarded is also remarkable because the competition for these NIH grants has increased substantially over the same time period due to a flattening of the NIH budget and an increasing number of applications.

Barth Syndrome Registry & Repository

One substantial hurdle faced by many clinical researchers is to find and enlist the support of patients affected by the disease being studied. This is especially true for rare disorders. Gaining access to patients can be a daunting task, and gathering extensive, longitudinal data on each patient can be next to impossible. This is one of the reasons rare disorders are often called “orphan” diseases and attract little research attention.

With Barth syndrome, we have a significant advantage in that we have taken the last few years and invested time, expertise and money to create and develop the Barth Syndrome Registry and Repository (BRR). The data and samples in our BRR are legally owned and controlled by BSF but housed and operated for us by a major educational and research institution which serves as the “warehouse” for clinical data about those with Barth syndrome (data reported by families, retrieved directly from medical records or collected during the clinics at our biennial international conferences). The BRR also contains biological samples, including DNA, various cell lines (lymphoblast lines made from blood samples and fibroblast lines created from skin samples) and a number of tissues from Barth syndrome (BTHS) patients such as hearts that have been removed during transplants and tissues that have been generously and selflessly donated at autopsy. Both the clinical data and the biological samples are extremely precious and are critical elements of our push toward more advanced clinical research and the development of therapies for Barth syndrome.

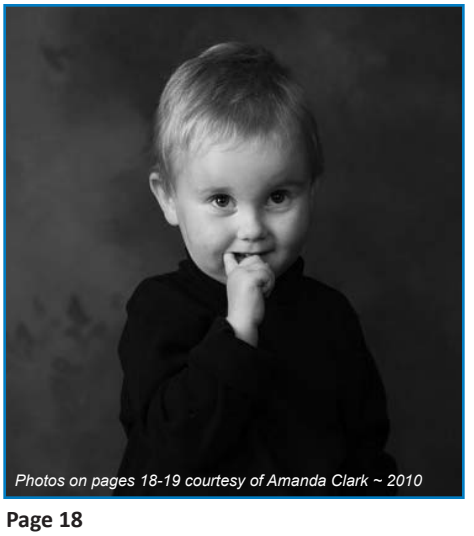
Several exciting developments have occurred recently that we want to share with you:

1. The BRR was transferred from the University of Florida (UF) to the Children’s Hospital Boston (CHB). UF was a good partner as we set up this critical resource, but the move to CHB provides some real benefits. In Boston, Dr. Carolyn Spencer continues to be involved, and she has gained the attention of a number of new subspecialists as well. She also has brought in Dr. Amy Roberts as an additional Principal Investigator. Dr. Roberts is the Director of the Cardiovascular Genetics Research Program at CHB and runs the hospital’s main registry for children with heart disease. Families, physicians and researchers who attended the July 2010 Barth Syndrome Conference had a chance to meet her and benefit from her expertise. Already, we have seen positive differences in focus and increasing momentum with the BRR, and we are excited about the progress.

2. Significant financial support for the BRR at CHB has been won through two grants from donors in the Boston area—an anonymous foundation and a generous individual, Christopher McKown. We are very grateful to these donors who appreciate the value of a comprehensive BRR to Barth syndrome research.

3. More IRB-approved clinical data was collected at the July 2010 Conference than ever before. “IRB-approved” means that the researchers gathering data have been approved by their own Institutional Review Board to conduct specific, carefully designed and executed research of the highest scientific quality, ensuring protection of the individuals who participate. Importantly, it also means that the clinical data can be used in peer-reviewed scientific articles.

The real measure of success of a registry and/or biorepository is what it produces, and we are beginning to see some results. A handful of scientists from around the world have applied to use BTHS lymphoblast and fibroblast cell lines and are conducting research on them currently. Researchers are beginning to request clinical data as well. Most of those who have approached the BRR are scientists BSF knows, but some are new, and that is especially exciting. We can see that our strategy of making access to critical data and samples easier is drawing in more researchers and facilitating their work!



Photos on pages 18-19 courtesy of Amanda Clark ~ 2010

Finances 2010

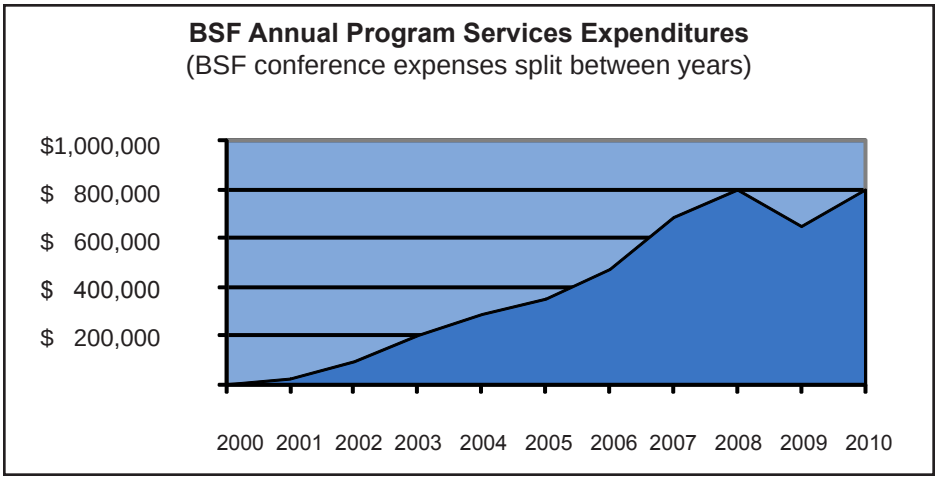
Like most charities, the Barth Syndrome Foundation (BSF) had a difficult year in 2010 where money was concerned. We ended the year with a deficit of $271,861 having raised $713,988 in contributions. On the positive side, we funded $253,894 in research grants and our most heavily attended International Barth Syndrome Conference at a cost of just over $200,000 including Poster stipends and speaker travel.

The Barth Syndrome Foundation, Inc. Statement of Activities For the Years Ended December 31			
	2010	2009	2008
Public Support and Revenue			
Contributions	$ 769,756	$ 694,771	$ 727,957
Other	$ 25,375	$ 43,592	$ 133,788
Total Support and Revenue	**$ 795,131**	**$ 738,363**	**$ 861,745**
Expenses			
Program Services	$ 902,379	$ 551,816	$ 894,904
Management and General	$ 131,225	$ 161,480	$ 160,139
Fund Raising	$ 33,388	$ 45,652	$ 11,582
Total Expenses	**$ 1,066,992**	**$ 758,948**	**$ 1,066,625**
Change in Net Assets	**$ (271,861)**	**$ (20,585)**	**$ (204,880)**
Net Assets - Beginning of Period	**$ 2,204,320**	**$ 2,224,905**	**$ 2,429,785**
Net Assets - End of Period	**$ 1,932,459**	**$ 2,204,320**	**$ 2,224,905**

What allows BSF to continue to fund our programs despite a shortfall in fund raising is what our accountants call our “Net Assets” — essentially prior year’s surplus funds saved and held in reserve for just such a year. We had $1,932,459 in Net Assets on our Balance Sheet at the end of 2010 which allows us to continue to fund critical science and medicine programs such as the Barth Syndrome Registry and Repository (BRR) and the special projects such as the development of the Barth mouse, in addition to the Research Grant program and the biennial Barth Syndrome Conference. Our annualized Program Expenditures have peaked at around $800,000.

(Cont’d on page 20)

(Cont'd from page 19)

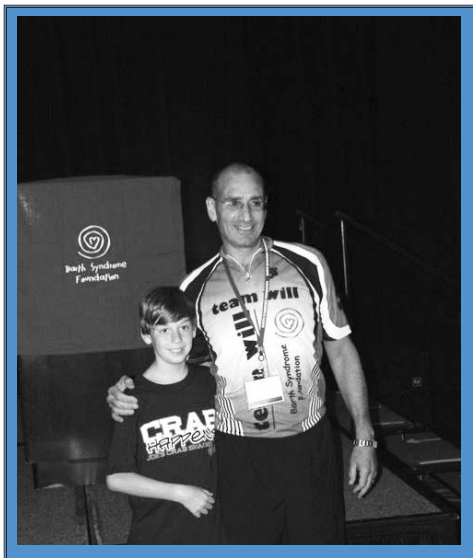


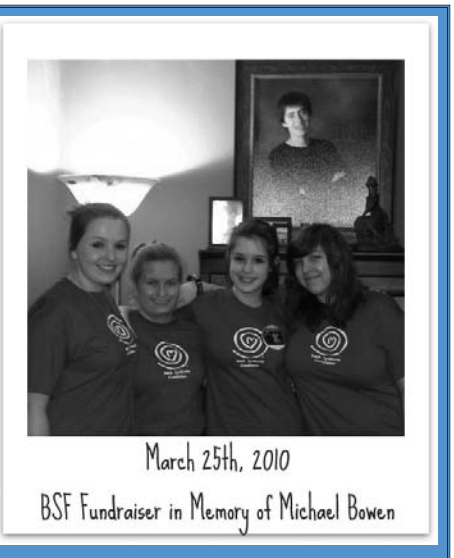
Still, it is imperative that all who believe in and support the work of BSF help us to find ways to raise additional money. All of our fund raising continues to come from friends and family members and from family foundations closely associated with them, from fund-raising efforts both large and small, and from donors whose gifts range from $5 to $100,000. In 2010, we received 874 donations from 571 individuals and institutions. The majority of our funds in 2010 came from the efforts of 13 Barth families, supplemented by a number of additional families who asked that gifts be made to BSF in memory of a loved one who had passed away—an incredible act of generosity in a time of great sadness.

BSF's Fund Raising Families - 2010	
McCurdy	Baffa
Buddemeyer	Oldewage
Sernel	Fairchild
Wilkins	Holly
Bowen	Higgins
Kugelmann	Brody
Osnos	

If the Barth Syndrome Foundation is to continue in our quest to understand the causes of Barth syndrome, insure the accurate diagnosis of every child affected, encourage the development of effective treatments and find a cure for this disorder, we need to add to this list of leaders! The McCurdys and Wilkins send out a simple letter each year to their friends and family explaining why they are devoted to our cause and asking that everyone include BSF in their charitable giving. The Kugelmann and Buddemeyer families sponsor golf tournaments and raise awareness and money under the sun in Florida. The Higgins family has held a fun-filled bowling tournament for years, giving their friends an extra excuse to give to BSF.

All these efforts are greatly appreciated on many levels. Not only do they garner important funds for our Foundation's work, but they also raise awareness of our little known and thus rarely diagnosed disease. So, as a result, our mission is advanced in several critical ways. Thank you to all who have been involved so far in any way, and thank you in advance to those who will join with even greater vigor this year.





Photos of 2010 fundraising events held in support of BSF.

(Photos courtesy of BSF ~ 2010)

2010 Donors

PAULA & WOODY VARNER FUND

Stars ($5,000+)
Yanney, Michael & Dr. Gail

Angels ($1,000 - $4,999)
Burmeister, Chuck & Marita
Wilkins, Sue & Dr. Mike

General ($50 - $999)
Allman, Peter & Maureen
Allman, Tom & Jane
Basler, Dr. Rod & Debbie
Beynon, Dave & Liz
Bingham, Dr. Dave & Kathy
Black, Bernice
Brehm, Russell & Louise (Center Associates, LLC)
Burmeister, Chuck & Marita
Campbell, Bob & Candy
Cheatham, Dr. John
Cheatham, Linda
Ditmann, Alice
Farrar, Doug & Shawn Seacrest Farrar
Firestone, Dave & Jane
Gelber, Dr. Ben & Elaine
Haessler, John & Nancy
Hayes, Dottie
Hedgecock, Norm & Debbie
Henricks, Dr. Bruce & Peg
Hille, Jim & Tina
Jensen, Tom & Marilyn
Kenyon, Barbara & Allan
Kiechel, Dr. Fred & Vivian
Lincoln Community Foundation
Linder, Dr. Max & Pat
Massengale, Dr. Martin & Ruth
Nelson, Scott & Teri
Norris, Dr. Mike & Mary
Olson, Loy & Julie
Osborne, Dr. Tom & Nancy
Otte, Rob & Carolyn
Pittenger, Jim & Julie
Raun, Robert & Eileen
Richards, Matt & Jessica
Roskens, Ronald & Lois
Roth, Dr. Bob & Marcia
Seaman, Andrew & Robyn Steely
Shamblin, David & Susan
Somers, Peter & Kristin
Sonderegger, Ted & Mary Ann
Stohs, Gene & Kris
Stuart, James & Susan (James Stuart Jr. & Susan Stuart Foundation)
Stuckey, Dennis & Nancy
Swanson, Mary
Tegt, Dr. Tom & Barb
The Kubly Family Foundation
Varner, Judy
Varner, Tom & Beth
Wiederspan, Mark & Jess
Wilkins, Dr. Jerry
Wilkins, Joanne
Wilkins, Marilyn
Wilkins, Dr. Lee & Kristi

Science & Medicine Fund

Stars ($5,000+)
Blumenthal, Richard & Cynthia
Lummis, Frederick (The Frill Foundation)
Malkin, Peter & Isabel (The Malkin Fund, Inc.)
Malkin, Laura & Scott (Scott D. Malkin 2005 Family Trust)
McCurdy, Steve & Kate
McKown, Christopher (Christopher J. McKown Charitable Gift Fund)
Russell, Dr. Paul
Sernel, Marc & Tracy

Science & Medicine Fund *(cont'd)*

Angels ($1,000 - $4,999)
Haines, Dr. Thomas
Kirkland & Ellis Foundation
Lummis, Ghent

General ($50 - $999)
Dudka, David
Cottell, George & Candace
Crabtree, John & Joyce
Dunagan, Kathlyn
Dunn, Mark & Anna
Dunn, Roland & Jean
Dunn, Stephen & Rosette
Guardabascio, Mary & Anna Pope
Harrigan, Timothy & Lori
Howe, George
Katuramu, Claire
Martin, Terrence & Ellen
Laperle, Rick & Ann Marie
O'Connor, Mary
O'Brien, Dennis (Dennis B. O'Brien Land Surveying)
Olson, Ken & Tina
Ouellette, Bob & Michelle
Ouellette, Randy & Tina
Schermerhorn, Mary
Szczepanek, Janelle
White, James & Lisa
Witzani, Gregor & Sonja

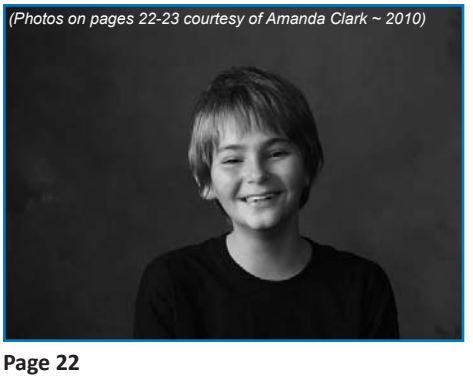
GENERAL FUND

Stars ($5,000+)
Barad, Seth & Amy
Buddemeyer, Randy & Friends (5th Annual Charity Golf Tournament)
Blumenthal, Richard & Cynthia
CB Richard Ellis
Communications Intl Inc.
Cusack, Tom & Carrie
Edward C. Johnson Fund
Janus Foundation
Lake City International Trucks, Inc.
Lummis, Marilyn
Lummis, William & Dossy
McKown, Christopher (Christopher J. McKown Charitable Gift Fund)
Osnos, Susan & Peter
Pierson, Allene & Dr. Robin
Pietrini, Andrew (The Lebensfeld Foundation)
Shapiro, Brenda
Vaisman, Natan & Beth

Angels ($1,000 - $4,999)
Alisberg, Andy & Susan
American Express Gift Matching
American Express PAC Match
Anonymous
Bailey, Clarke & Trish (The Patricia & Clarke Bailey Foundation, Inc.)
BSF Canada
Buddemeyer, Randy & Leslie
Cain, Rita & Jim
Cloud, Jody
Crowley, Gretchen & Peter
Davis, Martha (The Martha Davis & Richard Sayrs Family Fund)
Evoy, Sally & Larry
Fowler, David & Susan
Garry, Bobby & Leigh
Geary, Ann
Grose, Nora & Maddy
Hart, Dana
Hechinger, Nancy (Renaissance Charitable Foundation)
Hinchcliffe, Brian & Carol
Ingersoll, Ann
Isaac, Karen & Paul (Karen Cromer Isaac Fund)
Johnson, Malcolm & Deloris
Joyce, David & Jane
Kavetas, Suzanne (The Harry L. Kavetas Family Fund)
Kearns, Richard & Katherine
Kennett, Marie & Frank
Kirkland & Ellis Foundation
Kugelmann, Peter & Karen
Lake City International Trucks, Inc.
Lake City Idealease, LLC
Lummis, Marvin
Malkin, Eliza (Elizabeth Lummis Malkin Fund)
Malkin, Emily (Emily Mason Malkin Fund)
Malkin, Becky (Rebecca Swift Malkin Fund)
Mann, Allen & Rosa
McCormack, Joseph (Tides Foundation)
Neff, George & Elizabeth
Neviaser, James
Osnos, Suze & Peter
Phelps, Jim & Betty
Randolph, Dr. Peter & Helen
Schlapak, Gregor & Sonja
Schwartz, Marie & Mark
Shapiro, Earl & Brenda
Stoll, Ned & Cindy (Stoll & Stoll)
Taussig, Tim & Nancy (Tortimaki Foundation)
United Space Alliance
Winston, Laura

General ($50 - $999)
Abbonizio Contractors
Abbonizio, Leita
Adlam, Patty & Dr. Jim
Albert, Harold,
Allen, Tiffini & Kevin
America's Charities
Ameriprise Employee Gift Matching
Anderson, Tommy & Allanna
Andrews, David
Anonymous
Appell, Richard & Donna
Asaff, Ernie & Collette
Baffa, Ethel
Baffa, Ted & Rosemary
Bailey, Clarke & Trish (The Patricia & Clarke Bailey Foundation, Inc.)
Ballard, Stacey
Bancroft, Rev. Frank
Barth, Andrew & Allison
Bell, Roberta
Berens, Wayne & Harriet
Berland, Jonathan & Leslie
Berning, Mary Dell & Melvin
Berns, Scott
Berry, Carla & Rick
Berzins, Bob & Mara
Bialo, Kate & Ken
Bitterberg, Patricia
Black, Bernice
Bogert, Amy & Robert Baldwin
Bogert, Carroll
Bogert, Nicholas & Sally
Boschi, John & Anne
Bowen, Shelley & Michael
Bower, Scott & Karen
Bradley, Robert & Margaret
Brefka, Cornelia
Brennan, BJ
Brenner, Andy & Kathy
Brevard County Fire Rescue Dispatchers
Brody, D.W. & Tracy
Brown, Cary & Tracy
Browne, Lynne (Browne Family Charities)
Buddemeyer, Carl & Donna
Buddemeyer, David
Bullock, Betty
Buly, Lynne & Dr. Bob
Burmeister, Chuck & Marita
Burtis, Cleo & Jim
Butera, Jacquie & Donald Kyle
Butera, Thomas & Morlene
Callahan, William & Lynn
Campana, Owen
Carroll, Bob & Ann
Carroll, Brian & Randy
Carvalho, Maria Merlene
Castillo, Laura
Cavanna, Dino & Barbara
Cellino, Michael & Gina
Chandler, Ned & Brie
Chapin, Stephen & Deborah
Citta, Laura
Clark, Randy & Bob
Cohen, Chuck (Fabtrends International, Inc.)
Colagiuri, Ruth
Collazzi, J.J. & V.C.
Community Health Charities of California
Concannon, Maura & Dick (Concannon Family Foundation)
Conley, Joan & Stacey
Coogan, Susan
Cooper, James & Mary
Cox, Ray & Lou
Crabtree, John & Joyce
Cracchiolo, Jim & Marilyn
Crowley, Chuck & Diane
Cullison, James & Margaret
Cunniffe, Bernie & Rosemarie (Precision Engraving Group)
Cunniffe, Margaret (Sisters of St. Francis)
Damin, Michaela & Marco
Dannels, Richard & Therese
Davis, Beth & Friends
Day, P.T., Jane
Delaware Valley I.D. Associates, P.C.
Deshong, Howard & Diane
Dillworth Paxson
Dix, Lee & Glenda
Dixon, Julia
Doherty, Henry
Doherty, James & Kimberly
Donnalley, Jen
Drake, Bryan & Erin
Drake, Frank & Teresa
Duffy Real Estate, Inc.
Dunagan, Kathlyn
Dunn, Mark & Anna
Duran, Lisa
Egan, Thomas
Egueur, Madeleine
Elwood, Lynn & Rick
Emma, Alfred
Engel, Frederick & Ruby
Epstein, Paul & Leah
Evans, Robert & Beverly
Evans, Thomas & Sandra
ExecComm LLC
Fairchild, Julie & Dewayne
Farrell, Tim & Susan
Fedich, Allison
Fero, Douglas & Terry
Floyd, Ed & Julie
Flynn, Clare
Forsyth, John & Linnet Tse
Franklin, Peter & Dorothy
Fuller, Jim & Barbara
GRM Automotive
Gallagher, James & Mary Jane

(Photos on pages 22-23 courtesy of Amanda Clark ~ 2010)

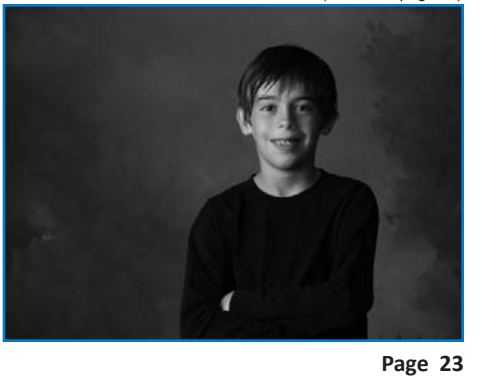


Gallaher, Carolyn
Garcia, Rudy & Linda
Garrity, Jon & Caroline
Gatheral, Jim
Geithner, Deborah & Peter
Gerfen, Henry
Gewitz, Dr. Michael & Judy
Glasgow, Merrill & Betsy
Glenn, William
Golden, Michael
Goldenberg, George & Arlene
Goldsmith Weiss Foundation
Gonnella, Tom & Jane
Gonzalez, Dr. Iris & Raul
Goodwin, Bill & Ginny
Gray, Melany & Dusty
Green, Mitchell & Susan
Greenberg, Dr. Miriam & Shifra
Greene, Rev. Dorothy
Gress, Emil & Natalie
Grossman, Dr. Harvey
Grzesiak, Lisa
Grzesiak, Richard & Eileen
Guerri, Robert
Gunderson, Roy & Jill
Gunther, Virginia & Tom
Guthorn, Robert & Marguerite
Halbur, Troy & Annette
Hales, Bryan
Hall, Dacey & Greg
Hall, James & Amie
Hall, Stuart & Stephanie
Halpern, Marc
Hansen, Christina
Harley, Rick & Elma Rhea
Harman, James & Merlene
Harris, Matthew
Harris, Nancy
Hartel, Charles
Hayes, Edward
Hazen, Liz & Ned
Hechler, Diana & David
Heller, Gerald & Sharon
Hepp, Timothy
Herr, Cabby
Heslin, Frank & Linda
Hiden, Jr., Bob & Ann
Hoffman, Laura
Hoffman, Michele & Binky
Holland, Mark & Diane
Holly, Greg & Keli
Holly, Peggy
Holman, Melinda & Frank
Hone, Susan & Chris
Hook, Stephen
Hope, Christine & Michael
Horton, Nancy
Howell, David & Patricia
Hubby, Pam
Hunt, Jeffrey & Catherine
Ingerman, Mark & Rebecca
Ingersoll, Ann
Ingersoll, Jared & Tina
Ingersoll, Paul & Mariana
Jaillet, Michael & Lizabeth
Jarvis, Rochelle
Jelco Enterprises Inc.
Jofre, Jaime
Joyce, Winifred
Kamph, Richard
Karofsky, Gale
Kearney, Patricia
Kelly, Raymond
Kelly's Security Service, Inc.
Kerr, Donald & Jean
Kim, Eugene
Kirkham, Ann & Collier
Kirtner, Holly
Kiwanis Club of Perry, Inc.
Kline, III, Talcott
Klockner, Dan & Nancy
Knapp, James & Maryanne
Knight, Jini & Rose Marie
Knopp, Cheryl
Kocher, Brenda
Krug, Thomas & Helen
Krynski, Steven & Regina
Kugelmann, Mike & Catherine
Kugelmann, Steve & Jan
Kuhl, Phillips & Karen
Latourette, John & Audrey
Layton, Alanna
Lehr Family Trust
Leon, Susan, Andy, Josh & Ali (The Leon/ Fredman Family)
Leone, Lewis & Claire
Liebenguth, Mouse
Lieblein, Carole
Lind, Michael
Lipson, Matthew
Liquidnet Double Down Program
Liscio, Liz & Mark
Livi, George & Jeanmarie
Lummis, Palmer & Mallory
Lupowitz, Kevin
Mahoney, John & Stephen
Mann, Allen & Rosa
Mann, John
Mann, Shelia & David
Mannes, Philippe & Florence
Mariani, Dominick & Catherine
Mariani, Michael & Christina
Marks, Cynthia & Michael
Mask, Harold & Gayle
Mayers, Bruce
Maynard, Dr. Ed & Lisa
McCarthy, Elizabeth
McCaughan, Elizabeth
McCaughan, Sean & Jeanette
McClintock, Cynthia & Mike
McCurdy, Mac & Ginny
McCurdy, Will
McEvily, Michael
McFee, Wendy & Jmel Wilson
McGrail, Sean
Meighan, Paula
Meny, Thomas
Merlino, Samuel & Patricia
Michaud, Steve & Yoko
Microsoft Matching Gifts Program
Miller, Kirt & Martha
Minniti, Sue
Misad, Marian
Mixter, Steve & Beth
Monetti, John & Rejane
Montanaro, Louis & Theresa
Morehouse, Susan & Clark
Morgenstern, Louise & Marc
Mulhern, Robert & Mary Kathleen
Nalley Lexus Roswell
Needham, James & Leslie
Neff, George & Elizabeth
Nentwig, Leo & Marilyn
Nersten, Timothy
Nicoll, Rev. Tom & Mary
Northern Kentucky Area Planning Commission
Northrup, Jan & Bruce
O'Brien, Justus, c/o Egon Zehnder International
O'Brien, John & Eileen
O'Connor, Mary
Odouard, Francois & Reshmi
Ohls, Mark & Carla
Olson, Richard & Sharon
Orlich, Charles
Osnos, Evan
Pagano, James & Mary Lou (Fully Involved Lawn Service)
Pahl, Carl & Jean
Paolizzi, Barbara
Papone, Aldo & Sandra
Pascoe, Richard
Pate, Deborah
Patil, Vinay
Pattee, Diane
Pearson, Inc.
Pepsico Foundation
Perez, Mr. & Mrs. Perez
Peterson, Carl & Andrea
Pfizer Foundation Matching Gifts Program
Phelps, Jim & Betty
Philip, Robert & Kathy
Phillips, Carolyn
Phillips, Kenneth & Margaret
Pierson, Allene & Dr. Robin
Pierson, Caroline
Pierson, Dick & Kitty
Pierson, Frank & Nancy
Pietrini, Andrew & Pam Mari
Pineault, Mitch
Plansky, Rita
Plumez, Jackie & Jean
Purcell, Jackie & Bob
Queenan, Jeri & Charlie
Rader, Carol
Rader, Stephanie
Ratliffe, David
Rey, Lisa
Roberts, John & Betty
Robinson, Francis & Sharon
Rodbell, Gary & Colette
Rodbell, Mitchell & Liz
Rogers, Cathy & Larry
Rothschild, Adam & Kathy
Rounsavill, Bruce & Rebecca
Russell, Dr. Nina & Tom Rubin
Russell, Trip & Sara
Russo, Bobby & Dottie
Salamone, Richard & Dolores
Sanborn, Noel & Virginia
Sandoval, Dr. Claudio
Sanford, Katherine & Colin
Scanlon, Erik (Real Time Lead Generation)
Schlame, Dr. Michael & Laili
Schlossberg, Martin (Castle Hill Apparel, Inc.)
Schreiber, Mary Ann & Ed
Schwendener, Paul & Barbara
Sedefian, Lynda
Segal, Anne
Segal, Heather & Mark
Sernel, Gordon & Sharon
Sherbany, Dr. Ariel
Sherer, Tony
Sherer, Peter & Marilu
Shields, William & Maryann
Shontz, David
Sims, Frank
Singer, Scott & Mary Jo
Skerratt, Mark & Rachel
Skinner, Adrienne
Sleeper, Emmy Lou
Smith, Judi & Jeff
Smith, Edward & Tina Marie
Sorrento, Nora
South Street Restaurant Group
Spackman, Randy & Drena Marie
Sports Unlimited, Inc.
Spotts, Dr. Jules
Spradin, Debra
Staab, Thomas
Stalter, Cathi
Standley, Burgess & Caroline
Stanton, Maria & Greg
Steinberg, Jack
Stenson, Martin & Marie
Stevenson, Robert & Sharon
Stewart, Michael & Barbara
Strain, Fraser & Donna
Strauss, Judith
Swain, Ginny & Bob
Taylor, Thomas & Patricia
Tedona, Sylvia
Telles, Michael & Linda
Thomas, Marion
Tides Foundation
Toth, Dr. Matt & Marilyn
Tulchin, Nora & David
Tunguz, Stefan & Julie
Usala Chapter 830
Utopia Communications, Inc.
Vaisman, Natan & Beth
Van Valen, Jeanne & Ed
Van Velan, Kera
VanBuren Brown, Missy
Vardell, Elizabeth & Jamie
Viebranz, Elaine
Vogt, Jerre
Volpi, Olga
Volpi, Vincent & Susan
Von Eckroth, Wulf
Votaw, Jebby
Waters, Marty & Helen
Welcome, Carolyn & Michael
Weltlich, Bob & Dodie
Weltich, Steve & Sharon
Wenglin, Dr. Barry & Barbara
Werth, Amanda & Tim (Wyatt Maksin Fund)
White, Johnny & Joyce
Wicker, Carol & Judy
Wilkins, John
Wilkov, Marcy
Williams, Kathleen & Suzi
Wong, Kityen
Woodcock, William & Barbara
Yocum, Ed & Lucy
Zeigner, Terry
Zierk, Gail & Tom
Zierk, Holly

TIME AND ADVICE

Abrahams, Esq., Beth Polner
Acehan, Dr. Devrim
Adhihetty, Dr. Peter
Agellon, Dr. Lou
Aikens, NCMA/CSR, Sharonda
Andersen, Harold
Anderson, Wayne & Suzie
Aprikyan, Dr. Andrew
Avery, RD, LD/N, CDE, Rhonda
Azar, Laura
Baffa, Kevin
Baffa, Rosemary
Barad, Seth
Bargo, PT, Jennifer
Barth, Dr. Peter G.
Bazinet, Dr. Richard
Bellamy, Amanda
Bennett, Dr. Michael
Benni, Dr. Paul
Beulow, Nancye
Beyer-Lead, Rick
Bogert, Nick
Bogert, Sally
Bolyard, RN, Audrey Anna
Bowen, Shelley
Bowron, Ann
Breitinger, PA, Petar
Brody, Tracy
Brown, Rebecca, Dir., Streetlight
Bruno, Ellen
Bryant, Dr. Randall M.
Buddemeyer, Leslie
Buddemeyer, Randy
Bullitt, Dr. Esther
Bungert, Dr. Jorg
Butera, MSW, CSW, MEd, Jaclyn
Byrne, Dr. Barry J.
Cade, Dr. Todd
Calhoun, Michael
Callahan, Lynn
Carboni, Dr. Michael
Carlson, Tony
Carney, Janet

(Cont'd on page 24)



TIME AND ADVICE *(cont'd)*

Carter, Kate Casidine
Cellino, Michael
Chase, Kevin
Christie, Dr. William W.
Clark, Amanda (Photographer)
Clay, Donna
Claypool, Dr. Steven M.
Corley, Marion
Cortez, Anne
Cox, Dr. Gerald F.
Crain, Carrie
Crawford, Lisa
Dale, Dr. David
Damin, Michaela
Dannels, Terry
Davies, Rob
Davis, Anastasia
Day, Dr. Jane
Develle, B.J.
DiMauro, Dr. Salvatore
Drinkwater, Paul
Duncan, CPA, Peter
Dunn, Anna
Dunn, Mark
Duran, Lisa
Elwood, Lynn
Epand, Dr. Richard
Epstein, Paul
Evans, Michael
Evans, Robert
Fairchild, Julie & Carl
Fan, Dr. Yuxin
Fei, Naomi
Fleischmann, Dr. Bernd
Flotte, Dr. Terry
Floyd, Julie
Flynn, Clare
Fortier, Jodie
Foxon, Kim
Fricker, Dr. Jay Frederick
Funke, Dr. Birgit
Gagnon, Lou Ann
Galbraith, Lois
Ganz, Doug
Gaudin, Herve
Gerszberg, Rich
Geva, Dr. Judith
Gill, Carrie
Gonzalez, Dr. Iris L.
Gottlieb, Dr. Eyal
Gravitt, Carolyn
Greenberg, Dr. Miriam L.
Groft, Dr. Steven
Haines, Dr. Thomas
Hammond, Dr. H. Kirk
Han, Dr. Xianlin
Hancock, PT, Lynn
Handisides, RN, Jill
Hardison, Haille & Jodi
Harley, Elma Rhea
Harrington, Hon. James
Hartford, Greg
Hartford, Larry
Hastings, Dr. Rob
Hatch, Dr. Grant M.
Hawkins, Lawton
He, Dr. Quan
Heal, Elisabeth
Henry, Anne
Henry, Dr. Susan
Higgins, John
Higgins, Liz
Hintze, Audrey
Hoffman, Laura
Holly, Keli
Holm, Ingrid
Hope, Chris
Hope, Michael
Hoppel, Dr. Charles
Houtkooper, Dr. Riekelt
Jacob, BS, Marni L.
Jailet, Michael & Lizabeth
Jefferies, Dr. John Lynn
Jensen, Dr. Robert E.
Jensen, Joanne
Jofre, Jaime
Johnson, Kristen
Johnson, Linda
Johnston, Dr. Jennifer J.
Joshi, Amit S.
Juico, MA, MEd, Eileen Q.
Kacinski, Debbie
Kainer, Daryl
Karp, Matt
Karp, Wendy
Kearns, Richard
Kelley, Dr. Richard I.
Kennedy, Yongyong
Kern, MGC, Rebecca L.
Khuchua, Dr. Zaza
Kiebish, Dr. Michael
King, OTR/L, Bobbie
King, Lynn
Kirwin, Susan
Koehler, Dr. Carla
Knopping, Jeff
Koehler, Dr. Carla
Kowalcyzk, Randy
Kreider, MHS, OTR/L, Consuelo
Kropp, Susan
Kugelmann, Dave
Kugelmann, Jan
Kugelmann, Lee
Kugelmann, Mike
Kugelmann, Sharon
Kugelmann, Steve
Kuijpers, Dr. Taco
Kulik, Dr. Willem
Laird, Doug
Lamoia, Michelle
Lane, Anna
Lawson, Lee Ann
Layton, Alanna
Leça, Dr. Ana
Lenaz, Dr. Giorgio
Levin, Dr. Gail
Lewin, Dr. Alfred S.
Lewis, Julie
Liphart, PT, DHSC, NCS, Jodi
Lipshultz, Dr. Steven E.
Lohmann, Jessica
Lowe, DHSc, PT, Jodi
Lucas, Kendal (Lucas Productions)
Lummis, Ghent
Lyall, Doug
Lynn, John
Madgett, Roberts, Marlowe, Jackson & Associates
Maisenbacher, MS, CGC, Melissa
Malhotra, Dr. Ashim
Mancino, Angelo
Mancino, Rosemary
Mann, Shelia
Mannella, Dr. Carmen
Manton, Annick
Margossian, Dr. Renee
Martin, Joy
Martins, Raquel
Maruno, Yuriko
Matthias, Linda
Mazzocco, Dr. Michele
McConaughy, Jim & Bev
McCormack, Joe
McCurdy, Eliza
McCurdy, Kate
McCurdy, Steve
McCurdy, Will
McMaster, Dr. Christopher
Miller, Cheryl
Miller, Travis
Mitchell, Jim
Mock, Kim
Monahan, Laurie
Monahan, Tom
Monetti, Kayleigh
Montenero, Theresa
Moore, Lorna
Moore, Nigel
Morava, Eva
Moreno-Quinn, Dr. Carol
Morris, Les
Morris, Travis
Murphy, Tony
Nackashi, Dr. John
Nixon, RN, Connie
Ntambi, Dr. James
Nunnari, Dr. Jodi
Nurse, Tom
Odouard, Francois
Odouard, Reshmi
Olson, Adam
Olson, Brandy
Olson, Dean
Olson, Maria
Olson, Richard
Olson, Sharon
Olson, Tina
Olson, Tom
Orstavik, Dr. Karen
Osnos, Susan
Pagano, Jim
Pagano, MaryLou
Patil, Vinay A.
Perkins, Phyllis
Pierson, Ali
Pilitowski, Bill & Colleen
Pittman, Jackie
Pruett, Debbie
Pu, Dr. William
Purcell, Robert & Jacqueline
Radosta, Lori
Randell, Amer
Redfearn, ARNP, Sharon
Reece, Bryce
Reiss, Dr. John
Ren, Dr. Mindong
Reppen, Heather
Reynolds, Stacey
Rigney, John
Rivkees, Dr. Scott A.
Rizzo, Dr. William
Roberts, Dr. Amy
Rodbell, Gary
Rodbell, Colette & Julia
Rosen, Alan
Rosen, Amy
Rosenshine, MA, MEd, Jonathan
Roubos, Mr. & Mrs.
Ruppshirts
Ryan, Jane
St. Amant, MS, Jay
Saidi, Dr. Arwa
Saroyan, Dr. John
Schantzen, Sandy
Schlame, Dr. Michael
Schrader, Cherie
Schroeder, Wallace & Alexis
Sedefian, Lynda
Segal, Heather
Segui, Damaris
Senthilnathan, Selvi
Sernel, Marc
Sexton, ARNP, Terry
Shenkman, Dr. Elizabeth
Sheppard, Jamie
Sherbany, Dr. Ariel
Sherman, Cathy
Sherwood, Dr. Geoff
Shih, Dr. Renata
Shirley, Alan & Denise
Shum, Bill & Ginny
Smith, Deborah
Smithson, Sarah
Smolski, Ed
Smoot, Dr. Leslie
Snyder, Floyd
Soustek, MS
Sparagna, Dr. Genevieve
Spencer, Dr. Carolyn T.
Stanford, RN, Dianne
Stehno-Bittel, Dr. Lisa
Steinberg, Jack
Steward, Dr. Colin
Stewart, Mr. & Mrs. Michael
Straits, Brian & Jan
Strauss, Dr. Arnold
Stundis, Linda
Sullivan, Melissa
Sutphin, Dr. Robert M.
Sydnor, Laurie
Taegtmeyer, Dr. Heinrich
Tarnopolsky, Dr. Mark
Taylor, Damani
Telles, Michael
Telles, Michelle
Thomas, Carla
Thompson, Angie
Thompson, Erin
Thorpe, Jeannette
Toniolo, Dr. Daniela
Toth, Dr. Matthew J.
Towbin, Dr. Jeffrey A.
Townsend, Esq., Colyn
Tsai-Goodman, Dr. Beverly
Tunguz, Stephen
Tweed, Scott
van der Riet, Hans
VanDuyne, PT, PCS, Jeannette
van Raam, Bram
Vancura, Dr. Ales
Varner, Judy
Vaz, Dr. Frederic M.
Vogt, Jerre
Vosgien, Don
Walker, Gena
Wallace, Dr. Douglas
Wanders, Dr. Ronald J. A.
Watson, MPT, Karin Colby
Weltlich, Dodie
Weltlich, Robert
Wenglin, Dr. Barry & Barbara
Whebble, Pam
White, Johnny & Joyce
White, Marty
Whitney, Scott (Whitney Media Productions)
Wicker, Carol
Wicker, Judy
Wiederspan, Jessica
Wiggins, Mark
Wilkins, Dr. Michael
Wilkins, John
Wilkins, Sue
Wilks, Carol
Williams, Kathy
Wise, Lisa
Wood, Robin
Wroe, Martha
Xu, Dr. Yang
Zaragoza, Dr. Michael
Zhang, Shali

(Photos courtesy of Amanda Clark ~ BSF 2010 Conference, July 2010, Orlando, FL.)

Governance

BSF is governed by our Board of Directors. Board members serve three-year terms and beginning in 2007 may not serve more than two consecutive terms. Annually, each Board member and officer signs a Conflict of Interest disclosure form and affirms in writing his/her obligation to protect the confidence of private information that BSF may acquire from families, physicians and researchers, as well as donors. Our Board members and officers and their terms are as follows:

Barth Syndrome Foundation Board of Directors

Name	Address	Position/Current Term End
Stephen B. McCurdy (Volunteer)	12 Carleon Avenue Larchmont, New York	Board Chairman Term Expiration 2013
Valerie M. Bowen (Staff)	205 Puckett Road Perry, Florida	President and Board Member Term Expiration 2012
Randy Buddemeyer (Volunteer)	1048 Paseo Del Rio NE St. Petersburg, Florida	Treasurer and Board Member Term Expiration 2011
Michaela Damin (Volunteer)	1 The Vikings Romsey, Hampshire United Kingdom	Board Member Term Expiration 2013
Stephen Kugelmann (Volunteer)	7145 Briar Oak Drive Merritt Island, Florida	Board Member Term Expiration 2012
Katherine R. McCurdy (Volunteer)	12 Carleon Avenue Larchmont, New York	Board Member Term Expiration 2011
Susan S. Osnos (Volunteer)	272 Round Hill Road Greenwich, Connecticut	Board Member Term Expiration 2012
Marcus E. Sernel (Volunteer)	201 S. Home Avenue Park Ridge, Illinois	Corporate Secretary and Board Member Term Expiration 2012
Susan V. Wilkins (Volunteer)	6219 Barbara Lane Lincoln, Nebraska	Board Member Term Expiration 2011

Barth Syndrome Foundation Executive Staff

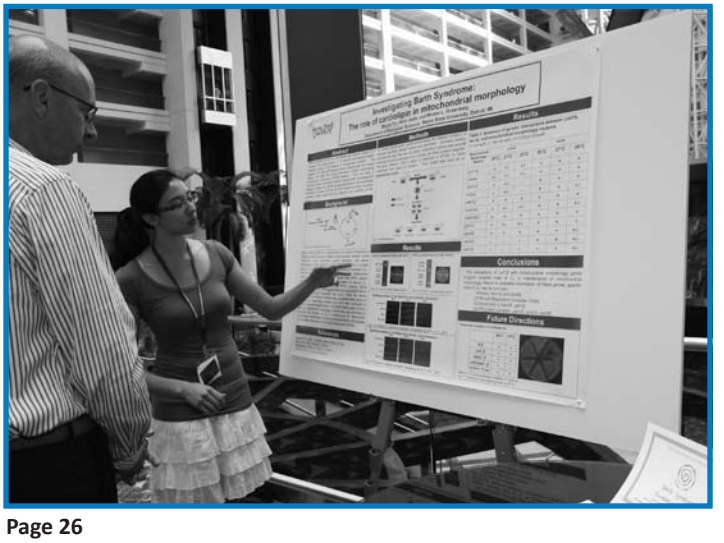
Name	Address	Position
Valerie M. Bowen	205 Puckett Road Perry, Florida	President
Matthew J. Toth, PhD	132 Creemer Avenue Iselin, New Jersey	Science Director
Lynda M. Sedefian	104 Stone Ridge Court Altamont, New York	Executive Assistant

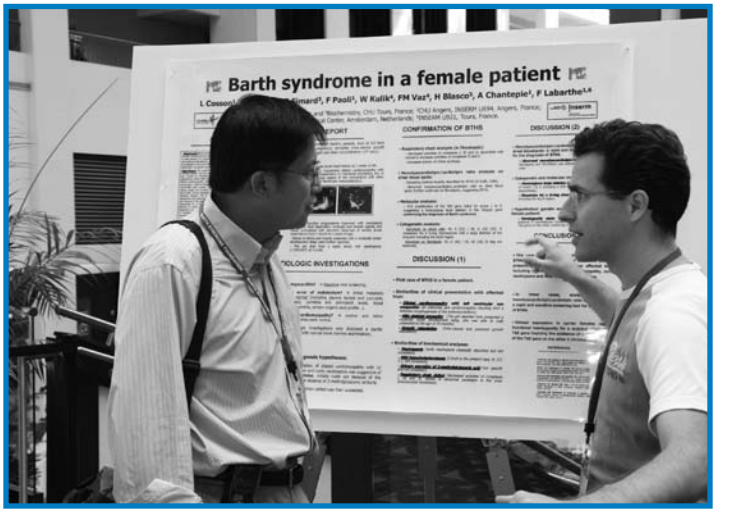
Scientific and Medical Advisory Board

The BSF Scientific and Medical Advisory Board (SMAB) is a world-class group of scientists and physicians with expertise in the diverse fields of research and the multiple systems affected by Barth syndrome (BTHS). These advisors review all research grant proposals and make their recommendations to the BSF Board which retains final grant approval authority. In addition, the SMAB advises on scientific and medical programs, participates in the biennial international conferences, and the SMAB clinicians offer consultations to our families and their physicians.

The Barth Syndrome Foundation Scientific and Medical Advisory Board

Richard I. Kelley, MD, PhD — *Chairman* Division of Metabolism Kennedy Krieger Institute Johns Hopkins University Baltimore, Maryland	Miriam L. Greenberg, PhD Biological Sciences Wayne State University Detroit, Michigan	Ronald J. A. Wanders, PhD Genetic Metabolic Diseases Academic Medical Center Amsterdam, The Netherlands
Peter G. Barth, MD, PhD — *Emeritus* Pediatric Neurology (retired) Emma Children's Hospital/AMC Amsterdam, The Netherlands	Grant M. Hatch, PhD Lipid Lipoprotein Research University of Manitoba Winnipeg, Canada	Katherine R. McCurdy, *ex-officio* Board Member Barth Syndrome Foundation Larchmont, New York
Barry J. Byrne, MD, PhD Pediatric Cardiology Shands Children's Hospital University of Florida Gainesville, Florida	Michael Schlame, MD Cell Biology & Anesthesiology NYU School of Medicine New York, New York	Matthew J. Toth, PhD, *ex-officio* Science Director Barth Syndrome Foundation Iselin, New Jersey
Gerald F. Cox, MD, PhD Clinical Genetics Children's Hospital Boston, Massachusetts Senior Medical Director Clinical Research, Genzyme Corp. Cambridge, Massachusetts	Colin G. Steward, FRCP, FRCPCH, PhD Pediatric Hematology Bristol Royal Hospital for Children Bristol, England	
Iris L. Gonzalez, PhD Molecular Diagnostics Lab *(retired)* A. I. DuPont Hospital for Children Wilmington, Delaware	Jeffrey A. Towbin, MD Pediatric Cardiology Cincinnati Children's Hospital Cincinnati, Ohio	





Barth Syndrome Registry and Repository

Barth Syndrome Registry and Repository Advisory Board	
Amy Roberts, MD — Co-Principal Investigator Cardiovascular Genetics Research Program, Children's Hospital Boston; Clinical Geneticist, Harvard Medical School, Boston, Massachusetts	Carolyn Spencer, MD — Co-Principal Investigator Pediatric Cardiology, East Carolina University, Greenville, North Carolina
Barry J. Byrne, MD, PhD Pediatric Cardiology, Shands Children's Hospital; University of Florida, Gainesville, Florida	Michael Schlame, MD Department of Anesthesiology, New York University School of Medicine, New York, New York
Gerald Cox, MD, PhD Clinical Genetics, Children's Hospital, Boston, MA; Senior Medical Director, Genzyme Corporation, Cambridge, Massachusetts	Colin G. Steward, FRCP, FRCPCH, PhD Pediatric Hematology, Royal Hospital for Children, Bristol, England
Richard I. Kelley, MD, PhD Division of Metabolism, Kennedy Krieger Institute; Johns Hopkins University, Baltimore, Maryland	Matthew J. Toth, PhD, *ex-officio* Science Director, Barth Syndrome Foundation, Iselin, New Jersey
Melissa Maisenbacher, MS, CGC, *ex-officio* Medical Data Abstraction, University of Florida College of Medicine, Gainesville, Florida	

(Photos on pages 26-27 courtesy of Cherie Schrader ~ BSF 2010 Conference, July 2010, Orlando, FL..)

Barth Syndrome Foundation, Inc. and International Affiliates

Barth Syndrome Foundation, Inc.
P.O. Box 618
Larchmont, New York 10538
Telephone: (850) 223-1128
E-mail: bsfinfo@barthsyndrome.org
Website: www.barthsyndrome.org

BSF International Affiliates

Barth Syndrome Trust (United Kingdom & Europe)
Michaela Damin, Chair
1 The Vikings
Romsey, Hampshire
S051 5RG
United Kingdom
Telephone: +44 (0)1794 518785
E-mail: info@barthsyndrome.org.uk
Website: www.barthsyndrome.org.uk

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

Barth Trust of South Africa
Jeannette Thorp, Chair
49 Abelia Road
Kloof, Pinetown
3610 Natal
South Africa
Telephone: 082-465-1965
E-mail: jthorpe@barthsyndrome.org
Website: www.barthsyndrome.org/South_Africa.html

Association Barth France
Florence Mannes, Chair
12, rue Lalo
75116 Paris
France
Telephone: +33 1 45 00 86 12
E-Mail: associationbarthfrance@orange.fr
Website:
http://barthfrance.com/Association%20Barth%20France.html

Featured are many of our Barth boys/young men together with their siblings (and a few dads who took advantage of the photo op) at BSF's 2010 International Scientific, Medical and Family Conference. (Photo courtesy of Amanda Clark ~ 2010.)

THE BARTH SYNDROME FOUNDATION, INC.
(A 501 (c) (3) Organization)

AUDITED FINANCIAL STATEMENTS

DECEMBER 31, 2010

SOUTHWEST PARK
ESTWOOD, MA 02090
EL: 781-407-0300
AX: 781-407-0303

GG&G
GRAY GRAY & GRAY, LLP
CERTIFIED PUBLIC ACCOUNTANTS
VISION • DIRECTION • SUCCESS
WWW.GGGCPAS.COM

INDEPENDENT AUDITORS' REPORT

Board of Directors
The Barth Syndrome Foundation, Inc.

We have audited the accompanying statements of financial position of **The Barth Syndrome Foundation, Inc.** (A 501 (c) (3) Organization) as of December 31, 2010 and 2009, and the related statements of activities, functional expenses, and cash flows – indirect method for the years then ended. These financial statements are the responsibility of the Foundation's management. Our responsibility is to express an opinion on these financial statements based on our audits.

We conducted our audits in accordance with auditing standards generally accepted in the United States of America. Those standards require that we plan and perform the audits to obtain reasonable assurance about whether the financial statements are free of material misstatements. An audit includes examining, on a test basis, evidence supporting the amounts and disclosures in the financial statements. An audit also includes assessing the accounting principles used and significant estimates made by management, as well as evaluating the overall financial statement presentation. We believe that our audits provide a reasonable basis for our opinion.

In our opinion, the financial statements referred to above present fairly, in all material respects, the financial position of **The Barth Syndrome Foundation, Inc.** as of December 31, 2010 and 2009, and the changes in net assets and its cash flows for the years then ended in conformity with accounting principles generally accepted in the United States of America.

GRAY, GRAY & GRAY, LLP

Gray, Gray & Gray, LLP

March 11, 2011

THE BARTH SYNDROME FOUNDATION, INC.
(A 501 (c) (3) Organization)

STATEMENTS OF FINANCIAL POSITION

ASSETS

	December 31, 2010	December 31, 2009
ASSETS		
Cash and cash equivalents	$ 538,826	$ 252,470
Investments	1,401,337	1,902,083
Accounts receivable	89,369	145,927
Prepaid expenses	2,969	4,755
TOTAL ASSETS	$ 2,032,501	$ 2,305,235

LIABILITIES AND NET ASSETS

LIABILITIES		
Accounts payable and accrued expenses	$ 23,062	$ 30,918
Grants payable	76,980	69,997
	100,042	100,915
NET ASSETS		
Unrestricted	1,063,759	1,436,375
Temporarily restricted	868,700	767,945
TOTAL NET ASSETS	1,932,459	2,204,320
TOTAL LIABILITIES AND NET ASSETS	$ 2,032,501	$ 2,305,235

The accompanying notes are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
(A 501 (c) (3) Organization)

STATEMENTS OF ACTIVITIES

YEAR ENDED DECEMBER 31, 2010 AND 2009

	2010			2009		
	Unrestricted	Temporarily Restricted	Total	Unrestricted	Temporarily Restricted	Total
PUBLIC SUPPORT AND REVENUE						
Contributions	$ 394,881	$ 259,875	$ 654,756	$ 338,751	$ 356,020	$ 694,771
Grant income	115,000	-	115,000	-	-	-
Interest income	26,121	-	26,121	59,232	-	59,232
Unrealized loss on investments	(746)	-	(746)	(15,640)	-	(15,640)
Net assets released from restrictions:						
Satisfaction of program restrictions	159,120	(159,120)	-	326,259	(326,259)	-
TOTAL PUBLIC SUPPORT AND REVENUE	694,376	100,755	795,131	708,602	29,761	738,363
EXPENSES						
Program services	902,379	-	902,379	551,816	-	551,816
Management and general	131,225	-	131,225	161,480	-	161,480
Fundraising	33,388	-	33,388	45,652	-	45,652
TOTAL EXPENSES	1,066,992	-	1,066,992	758,948	-	758,948
CHANGES IN NET ASSETS	(372,616)	100,755	(271,861)	(50,346)	29,761	(20,585)
NET ASSETS AT BEGINNING OF YEAR	1,436,375	767,945	2,204,320	1,486,721	738,184	2,224,905
NET ASSETS AT END OF YEAR	$ 1,063,759	$ 868,700	$ 1,932,459	$ 1,436,375	$ 767,945	$ 2,204,320

The accompanying notes are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
(A 501 (c) (3) Organization)

STATEMENT OF FUNCTIONAL EXPENSES

YEAR ENDED DECEMBER 31, 2010

	Program Services	Management and General	Fundraising	Total
Salaries	$ 223,569	$ 60,190	$ 24,817	$ 308,576
Payroll taxes and benefits	52,915	13,419	3,705	70,039
TOTAL PERSONNEL SERVICES	276,484	73,609	28,522	378,615
Research grants	277,968	-	-	277,968
Professional	95,089	36,376	-	131,465
Telephone	8,010	3,055	-	11,065
Office expense	35,342	10,374	3,237	48,953
Printing and publications	9,433	118	-	9,551
Dues and fees	9,276	4,019	1,155	14,450
Transportation	69,337	144	54	69,535
Insurance	-	3,530	-	3,530
Meals	80,561	-	-	80,561
Audio visual expense	35,544	-	-	35,544
Miscellaneous	5,335	-	420	5,755
TOTAL EXPENSES	$ 902,379	$ 131,225	$ 33,388	$ 1,066,992

The accompanying notes are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
(A 501 (c) (3) Organization)

STATEMENT OF FUNCTIONAL EXPENSES

YEAR ENDED DECEMBER 31, 2009

	Program Services	Management and General	Fundraising	Total
Salaries	$ 225,700	$ 74,363	$ 36,176	$ 336,239
Payroll taxes and benefits	41,133	17,822	7,737	66,692
TOTAL PERSONNEL SERVICES	266,833	92,185	43,913	402,931
Research grants	144,687	-	-	144,687
Professional	67,870	40,164	-	108,034
Telephone	10,478	4,193	-	14,671
Office expense	11,715	11,266	-	22,981
Printing and publications	3,354	-	-	3,354
Dues and fees	4,841	6,388	1,000	12,229
Transportation	42,038	4,152	739	46,929
Insurance	-	3,132	-	3,132
TOTAL EXPENSES	$ 551,816	$ 161,480	$ 45,652	$ 758,948

The accompanying notes are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
(A 501 (c) (3) Organization)

STATEMENTS OF CASH FLOWS - INDIRECT METHOD

	Year Ended December 31, 2010	2009
CASH FLOWS FROM OPERATING ACTIVITIES		
Changes in net assets	$ (271,861)	$ (20,585)
Adjustments to reconcile changes in net assets to net cash (used) by operating activities:		
Unrealized loss on investments	746	15,640
Change in operating asset and liabilities:		
Accounts receivable	56,558	(138,692)
Unconditional promises to give	-	125,000
Prepaid expenses	1,786	(4,043)
Accounts payable and accrued expenses	(7,856)	15,337
Grants payable	6,983	(64,846)
NET CASH (USED) BY OPERATING ACTIVITIES	(213,644)	(72,189)
CASH FLOWS FROM INVESTING ACTIVITIES		
Acquisition of investments	(100,000)	(200,000)
Proceeds from redemption of investments	600,000	-
NET CASH PROVIDED (USED) BY INVESTING ACTIVITIES	500,000	(200,000)
INCREASE (DECREASE) IN CASH AND CASH EQUIVALENTS	286,356	(272,189)
CASH AND CASH EQUIVALENTS AT BEGINNING OF YEAR	252,470	524,659
CASH AND CASH EQUIVALENTS AT END OF YEAR	$ 538,826	$ 252,470

The accompanying notes are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
(A 501 (c) (3) Organization)

NOTES TO FINANCIAL STATEMENTS

DECEMBER 31, 2010

NOTE 1 – BUSINESS

Organization – The Barth Syndrome Foundation, Inc. (the "Foundation") is a not-for-profit organization incorporated under the laws of the state of Delaware on September 8, 2000 to be operated for the following purposes: a) to support and educate families with children suffering from Barth Syndrome; b) to fund and facilitate research addressing the causes, diagnosis, treatment, and cure of Barth Syndrome; and c) to raise physician awareness regarding Barth Syndrome (the "Syndrome").

NOTE 2 – SIGNIFICANT ACCOUNTING POLICIES

Financial Statement Presentation – The accompanying financial statements have been prepared on the accrual basis of accounting in accordance with accounting principles generally accepted in the United States of America. The presentation follows the recommendation of the Financial Accounting Standards Board under which the Foundation is required to report information regarding its financial position and activities to three classes of net assets:

- *Unrestricted* – Represents all activity without donor imposed restrictions.
- *Temporarily Restricted* – Relates to contributions of cash and other assets with donor stipulations that make clear the assets' restrictions, either due to a program nature or by passage of time.
- *Permanently Restricted* – Relates to contributions of cash and other assets whereby the assets must remain intact due to restrictions placed by the donor. The Foundation had no permanently restricted net assets at December 31, 2010 and 2009.

Contributions – Contributions received are recorded as unrestricted or temporarily restricted support depending on the existence and/or nature of any donor restrictions.

Support that is restricted by the donor is reported as an increase in unrestricted net assets if the restriction expires in the reporting period in which the support is recognized. All other donor-restricted support is reported as an increase in temporarily restricted net assets, depending on the nature of the restriction. When a restriction expires (that is, when a stipulated time restriction ends or purpose restriction is accomplished), temporarily restricted net assets are reclassified to unrestricted net assets and reported in the statement of activities as net assets released from restriction.

Cash and Cash Equivalents – Cash and cash equivalents include cash on hand and money market funds which have original maturities of three months or less. The Foundation maintains its cash and money market accounts at institutions they consider to be credit worthy. All cash and money market accounts and CD's held by the Foundation are insured by the Federal Deposit Insurance Corporation (FDIC) or Securities Investor's Protection Corporation (SIPC) at specific limits. During the course of the normal business cycle the Foundation may, at times, maintain cash and cash equivalent balances in excess of the FDIC and SIPC insurance limits. Cash and cash equivalents in excess of insurance limits amounted to $127,864 at December 31, 2010.

NOTE 2 – SIGNIFICANT ACCOUNTING POLICIES (CONTINUED)

Donated Assets – Donated marketable securities and other non-cash donations are recorded as contributions at their estimated fair values at the date of donation.

Investments – All investments are held in certificates of deposit and are measured at fair value in the statements of financial position. Unrealized gains or losses are included in the changes in net assets. Investment income is reported net of brokerage fees and commissions. Investment transactions are recorded on a trade date basis.

Financial Accounting Standards Board ("FASB") issued an interpretation, "*Fair Value Measurements*", which defines fair value, establishes a framework for measuring fair value, and expands disclosures about fair value measurements. Fair value is the price that would be received to sell an asset or paid to transfer a liability in an orderly transaction between market participants at the measurement date. A fair value measurement assumes that the transaction to sell the asset or transfer the liability occurs in the principal market for the asset or liability or, in the absence of a principal market, the most advantageous market. Valuation techniques that are consistent with the market, income or cost approach, as specified by FASB, are used to measure fair value. The fair value hierarchy prioritizes the inputs to valuation techniques used to measure fair value into three broad levels as follows:

Level 1 – inputs are quoted prices (unadjusted) in active markets for identical assets or liabilities the Foundation has the ability to access.

Level 2 – inputs are inputs (other than quoted prices included within Level 1) that are observable for the asset or liability, either directly or indirectly.

Level 3 – are unobservable inputs for the asset or liability and rely on management's own assumptions about the assumptions that market participants would use in pricing the asset or liability. (The unobservable inputs should be developed based on the best information available in the circumstances and may include the Foundation's own data).

Accounts Receivable and Allowance for Doubtful Accounts – Receivables are recorded at their estimated net realizable value. The Foundation records an allowance for estimated accounts receivable in an amount approximating anticipated losses. Individual uncollectible receivables are written off against the allowance when collection of the individual receivable appears doubtful. At December 31, 2010 and 2009, management determined that no allowance for doubtful accounts was required.

Capitalization Policies – Items of property and equipment with an individual cost in excess of $5,000 are capitalized at cost. Routine maintenance and repair costs and leasehold improvements, which do not materially extend the estimated useful lives of property and equipment, are expensed as incurred.

NOTE 2 – SIGNIFICANT ACCOUNTING POLICIES (CONTINUED)

Concentrations of Credit Risk – Financial instruments which potentially subject the Foundation to concentrations of credit risk consist principally of money market accounts, investments and accounts receivable. Concentrations of credit risk with respect to accounts receivable is limited due to the majority of the balances are contribution commitments due from board members, BSF affiliates, or companies and individuals associated with the board members.

Grant income – Grants are awarded to the Foundation primarily by private foundations and other non-profit organizations. The grants are deemed to be earned and reported as revenues when expenditures are incurred in compliance with the specific grant restrictions.

Expense Allocation – The costs of providing various programs and other activities have been summarized on a functional basis in the statements of activities and in the statements of functional expenses. Accordingly, certain costs have been allocated among the programs and supporting services benefited. Management and general expenses include those expenses that are not directly identifiable with any specific programs or fund raising activities but provide for the overall support and direction of the Foundation.

Income Taxes – The Foundation is a tax-exempt organization under Section 501 (c) (3) of the Internal Revenue Code and has not been designated as a private foundation.

The Foundation is required to recognize the financial statement impact of a tax position unless it is more likely than not that the position will be sustained upon examination. Any interest and penalties recognized associated with a tax position would be classified as current in the Foundation's financial statements.

Currently, the 2007, 2008, and 2009 tax years are open and subject to examination by the Internal Revenue Service and in various states that the Foundation is registered. However, the Foundation is not currently under audit nor has the Foundation been contacted by any of these jurisdictions.

Based on the evaluation of the Foundation's tax positions, management believes all positions taken would be upheld under an examination. Therefore, no provision for the effects of uncertain tax positions has been recorded for the year ended December 31, 2010.

Use of Estimates – The preparation of financial statements in conformity with accounting principles generally accepted in the United States of America requires management to make estimates and assumptions that affect certain reported amounts and disclosures. Accordingly, results could differ from those estimates.

NOTE 3 – INVESTMENTS

Investments consist of the following at December 31, 2010:

	Cost	Fair Value	Quoted Price Inputs (Level 1)
Certificates of Deposit	$ 1,400,070	$ 1,401,337	$ 1,401,337

Investments consist of the following at December 31, 2009:

	Cost	Fair Value	Quoted Price Inputs (Level 1)
Certificates of Deposit	$ 1,900,095	$ 1,902,083	$ 1,902,083

NOTE 4 – TEMPORARILY RESTRICTED NET ASSETS

Temporarily restricted net assets consist of the following as of December 31:

	2010			
	Balance 1/1/2010	Contributions	Released from Restrictions	Balance 12/31/2010
Program Restrictions:				
Paula & Woody Varner Science and Medical Fund	$ 146,696	$ 15,735	$ (1,461)	$ 160,970
Science and Medical Fund	621,249	244,140	(157,659)	707,730
Total	$ 767,945	$ 259,875	$ (159,120)	$ 868,700

NOTE 4 – TEMPORARILY RESTRICTED NET ASSETS (CONTINUED)

	2009			
	Balance 1/1/2009	Contributions	Released from Restrictions	Balance 12/31/2009
Program Restrictions:				
Paula & Woody Varner Science and Medical Fund	$ 168,094	$ 21,240	$ (42,638)	$ 146,696
Science and Medical Fund	445,090	334,780	(158,621)	621,249
Total program restrictions	613,184	356,020	(201,259)	767,945
Time restrictions	125,000	-	(125,000)	-
Total	$ 738,184	$ 356,020	$ (326,259)	$ 767,945

NOTE 5 – COMMITMENTS

Grants payable as of December 31, 2010 and 2009 consists of amounts awarded, but not paid, of $76,980 and $69,997, respectively. All are due to be paid within twelve months.

The Foundation is committed to research grants awarded subsequent to year end of approximately $308,000, that are payable in 2011 and 2012.

NOTE 6 – INTERNATIONAL CONFERENCE

The Foundation holds an international conference every two years and the related costs are expensed in the year incurred. The most recent conference was held in July 2010. The conference brings together doctors and scientists involved in the many aspects of the Syndrome to discuss the latest underlying scientific developments and clinical insights. In addition, the conference allows families to obtain the latest information relating to the Syndrome and consult with medical experts from around the world. The families are also provided the opportunity to provide important clinical data and biological samples to the Barth Syndrome Medical Database and Biorepository.

NOTE 7 – DONATED SERVICES

Many individuals volunteer their time and perform a variety of tasks that assist the Foundation with specific programs, campaign solicitation, and various committee assignments. No value has been assigned to these volunteer services, as the criteria for recognition under generally accepted accounting principles have not been satisfied.

NOTE 8 – SUBSEQUENT EVENTS

The Company has evaluated subsequent events through March 11, 2011, the date which the financial statements were available to be issued. There were no events noted that required disclosure in these financial statements.

ATTACHMENT A

SOUTHWEST PARK
ESTWOOD, MA 02090
EL: 781-407-0300
AX: 781-407-0303

GG&G
GRAY GRAY & GRAY, LLP
CERTIFIED PUBLIC ACCOUNTANTS
VISION • DIRECTION • SUCCESS
WWW.GGGCPAS.COM

November 10, 2010

Mr. Stephen B. McCurdy
The Barth Syndrome Foundation, Inc.
P. O. Box 618
Larchmont, NY 10538

Dear Mr. McCurdy:

This letter is being written to confirm and specify the terms of our engagement and to clarify the nature and extent of the accounting services we will provide for December 31, 2010.

We will audit the statements of financial position of **The Barth Syndrome Foundation, Inc.** as of December 31, 2010 and 2009, and the related statements of activities, and cash flows for the year then ended.

The objective of our audits is the expression of an opinion about whether your financial statements are fairly presented, in all material respects, in conformity with accounting principles generally accepted in the United States of America. Our audits will be made in accordance with auditing standards generally accepted in the United States of America and will include tests of your accounting records and other procedures we consider necessary to enable us to express an unqualified opinion that your financial statements are fairly presented, in all material respects, in conformity with accounting principles generally accepted in the United States of America. If our opinion is other than unqualified, we will fully discuss the reasons with you in advance. If, for any reason, we are unable to complete the audits or are unable to form or have not formed an opinion, we may decline to express an opinion or to issue a report as a result of this engagement.

Our procedures will include tests of documentary evidence supporting the transactions recorded in the accounts, tests of the physical existence of inventories, and direct confirmation of receivables and other assets and liabilities by correspondence with selected customers, creditors, and banks. We will request written representations from your attorneys as part of the engagement, and they may bill you for responding to this inquiry. At the conclusion of our audits, we will also request certain written representations from you about the financial statements and related matters.

An audit includes examining on a test basis, evidence supporting the amounts and disclosures in the financial statements. Our audits will involve judgment about the number of transactions to be examined and the areas to be tested. Also we will plan and perform the audits so that we may obtain reasonable assurance about whether the financial statements are free of material misstatement, whether caused from (1) errors, (2) fraudulent financial reporting, (3) misappropriation of assets, or (4) violations of laws or governmental regulations that are attributable to the Organization or to acts by management or employees acting on behalf of the Organization.

Because an audit is designed to provide reasonable, but not absolute, assurance and because we will not perform a detailed examination of all transactions, there is a risk that material misstatements may exist and not be detected by us. In addition, an audit is not designed to detect immaterial misstatements or violations of laws or governmental regulations that do not have a direct and material effect on the financial statements. However, we will inform you of any material errors that come to our attention, and we will inform you of any fraudulent financial reporting or misappropriation of assets that comes to our attention. We will also inform you of any violations of laws or governmental regulations that come to our attention, unless clearly inconsequential. Our responsibility as auditors is limited to the period covered by our audit and does not extend to any later periods for which we are not engaged as auditors.

We understand that you will provide us with the basic information required for our audit and that you are responsible for the accuracy and completeness of that information. We will advise you about appropriate accounting principles and their application and will assist in the preparation of your financial statements, but the responsibility for the financial statements remains with you. You and the Organization's management are responsible for maintaining adequate records, selecting and applying accounting principles, and safeguarding assets. You and the Organization's management are responsible for establishing and maintaining a sound system of internal control and a culture which promotes the integrity and accuracy of the Organization's financial books and records. We believe this is the best means of preventing or detecting theft, embezzlement, defalcations, illegal acts, errors, fraudulent financial reporting, and misappropriation of assets. If you have any concerns relating to these matters we expect you will speak candidly with us about them. We may be able to assist you in these areas by designing a special engagement specifically to address these matters.

We understand that your employees will prepare all cash, accounts receivable, accounts payable, and other confirmations we request and they will prepare schedules, as requested and locate supporting documents to minimize the time required by our auditors.

Our audit will include obtaining an understanding of the Organization and its environment, including internal control, sufficient to assess the risks of material misstatement of the financial statements and to design the nature, timing, and extent of further audit procedures. An audit is not designed to provide assurance on internal control or to identify deficiencies in internal control. However, during the audits, we will communicate to you and those charged with governance of internal control related matters that are required to be communicated under professional standards.

Management is responsible for adjusting the financial statements to correct material misstatements and for affirming to the auditors in the representation letter that the effects of any uncorrected misstatements brought to its attention by the auditors are immaterial, both individually and in the aggregate, to the financial statements taken as a whole.

As part of our engagement, we may propose standard, adjusting, or correcting journal entries to your financial statements. You are responsible for reviewing the entries and understanding the nature of any proposed entries and the impact they have on the financial statements. Further, you are responsible for designating a qualified management-level individual to be responsible and accountable for overseeing these services.

You are responsible for the design and implementation of programs and controls to prevent and detect fraud, and for informing us about all known or suspected fraud affecting the Organization involving (a) management, (b) employees who have significant roles in internal control and (c) others where the fraud could have a material effect on the financial statements. You are also responsible for informing us of your knowledge of any allegations of fraud or suspected fraud affecting the Organization received in communications from employees, former employees, regulators or others. In addition, you are responsible for identifying and ensuring that the Organization complies with applicable laws and regulations.

You are also responsible for management decisions and functions; for designating an individual with suitable skill, knowledge, or experience to oversee the bookkeeping, tax and any other nonattest services we provide; and for evaluating the adequacy and results of those services and accepting responsibility for them. We, in our sole professional judgment, reserve the right to refuse to do any procedures or take any action that could be construed as making management decisions or performing management functions. We will advise management with regard to tax positions taken in the preparation of the tax return, but management must make all decisions with regard to those matters.

We will not perform management functions or make management decisions on behalf of the Organization. However, we will provide advice and recommendations to assist management in performing its functions and making decisions.

We will prepare the Organization's federal (IRS Form 990) tax return for the year ended December 31, 2010. We will examine when appropriate, assessment notices relative to these returns, advise on tax matters generally, and discuss any matters concerning your taxes with the taxing authorities. Our advice concerning tax matters will relate to the proper tax treatment of transactions already completed and will depend upon the accuracy and completeness of the records provided to us. We will advise you with regard to tax positions taken in the preparation of the tax returns, but the responsibility remains with you. We may require a separate engagement letter for tax planning engagements and/or tax representation matters.

In the event we are requested or authorized by you or required by government regulation, subpoena, or other legal process to produce our working papers or our personnel as witnesses with respect to our engagement for you, you will, so long as we are not a party of the proceeding in which the information is sought, reimburse us for our professional time and expenses, as well as the fees and expenses of our counsel, incurred in responding to such a request.

Michael L. Cecere is the engagement partner and is responsible for supervising the engagement and signing the report or authorizing another individual to sign it.

GG&G
GRAY GRAY & GRAY, LLP

You agree to pay a fee of $8,250 for Gray, Gray & Gray's services, plus travel and other out-of- pocket costs. Our invoices for these fees will be rendered as work progresses and are payable on presentation. In accordance with our firm policies, work may be suspended if your account becomes overdue and will not be resumed until satisfactory arrangements for payment can be made. Late fees of 1 ¼% (15% per annum) will be charged on balances more than 60 days overdue. We reserve the right to withdraw from this engagement without issuing a report if we are not paid the full amount we are owed, or if we find that we have reason to question management's truthfulness or the integrity of the Organization's books and records.

The fee quoted above assumes the level of client assistance as described in our client preparation request letter sent under separate cover and the assumption that unexpected circumstances will not be encountered during the engagement.

By signing this engagement letter, you agree that our liability from this engagement shall be limited to the lesser of any actual damages which may have been caused by our acts or omissions or the amount of the fees which you pay for these services. We may agree to increase the limit of our liability in consideration of payment by client of additional monetary and other consideration. Please contact us if you wish to discuss this further.

Bear in mind that even though we may prepare the financial statements, the representation (i.e., words and numbers) in them are your representation since they are based on your transactions. We rely on what you tell us to be completely truthful. By signing this engagement letter you agree to indemnify us and hold us harmless from any liability and costs arising from knowing misrepresentations of management.

Should any questions arise as to the quality or timeliness of our services, we ask that you call such matters to our attention promptly. By signing this letter, we ask that you agree to submit any such dispute which is not resolved in that fashion; first, to voluntary, non-binding mediation before the American Arbitration Association, and that you will refrain from instituting legal action unless such mediation is exhausted without a resolution of the dispute.

With regard to the electronic dissemination of audited financial statements, including financial statements published electronically on your website, you understand that electronic sites are a means to distribute information and, therefore, we are not required to read the information contained in these sites or to consider the consistency of other information in the electronic site with the original document.

During the course of our engagement, you may provide us with certain documents and records. It is your responsibility to retain the original or a copy of the records and not our responsibility to retain them on your behalf.

It is our policy to keep workpapers, including copies of tax returns, relating to this engagement for seven (7) years on site. Thereafter, whenever appropriate, these records will be destroyed or stored off-site. It is your responsibility to retain and protect your records for possible use, including potential examination by any government or regulatory agencies.

GRAY GRAY & GRAY, LLP

In the future, you may decide that you need the services of an in-house controller. At that time, we can assist you in the selection of a qualified individual. If you find that because of our knowledge of your business, you wish to hire any Gray, Gray & Gray employee you will be charged a recruiting fee. This fee will be twenty percent of the annual salary offered to our employee, to compensate our firm for the loss of a valued employee.

While performing this engagement, we may deem it appropriate and in your best interest to involve a third-party service provider. In such instance, be assured we will be responsible for the work performed by this service provider and will use reasonable care to assure it has appropriate safeguards and procedures in place to protect your confidential information.

Several technical accounting and auditing words and phrases have been used herein. We presume you to understand their meaning or that you will notify us otherwise so that we can furnish appropriate explanations.

By signing this engagement letter you agree that any suit for enforcement of this Agreement may be brought in the Courts of the Commonwealth of Massachusetts or any Federal Court sitting therein and consent to the exclusive jurisdiction of such court. In addition, you hereby waive any objection that you may now or hereafter have to the venue of any such suit or any such court or based on such suit having been brought in any inconvenient forum. This Agreement and all rights and obligations hereunder, including matters of construction, validity, and performance, shall be governed by the laws of the Commonwealth of Massachusetts.

If this account is placed for collection, you agree to pay all costs and reasonable attorney's fees associated with the collection of any past due balance.

We appreciate the opportunity to be of service to you and believe this letter accurately summarizes the significant terms of our engagement. If you have any questions, please let us know. If you agree with the terms of our engagement as described in this letter, please sign the enclosed copy, and return it to us.

Very truly yours,

Gray, Gray & Gray, LLP

MLC:ptc

The services described in the foregoing letter are in accordance with our requirements. The terms described in the letter are acceptable to us and hereby agreed to.

Signature: _____
Title: _____
Date: _____

061580001210.ef

The Barth Syndrome Foundation has been accredited by the Better Business Bureau, meeting all accreditation standards.

The Barth Syndrome Foundation is a member of the National Health Council, improving the health of all people, particularly those with chronic diseases and/or disabilities. BSF abides by all 42 of NHC's best practices.

The Barth Syndrome Foundation is a member of the Genetic Alliance.

2010 Barth Syndrome Foundation

Photo courtesy of Amanda Clark ~ 2010