Barth Syndrome Foundation

Barth Syndrome Foundation®

2007 Annual Report

2007 Barth Syndrome Foundation Annual Report

To Our Major Donors:

Every year at this time, I have the pleasure of summing up our community's accomplishments in the previous year. 2007 again was a year when our dedicated volunteers, faithful contributors, caring physicians and brilliant researchers have created and accomplished much. This annual report reflects their triumph of progress and hope.

Most of the people on the front lines of these efforts are parents who have a very personal reason for volunteering. But in our determination to shape BSF into a powerful and permanent ally in the fight to save our children's lives, one thing keeps rising up to distract our attention and challenge our hopes; the fear that we may not do enough, fast enough to save our own child. Fear is a powerful thing. It can be highly motivating. But at times it can also be overwhelming. It can become a black, nauseating, and paralyzing demon in its threat to take from us those whom we love the most… and would gladly give our own lives to protect.

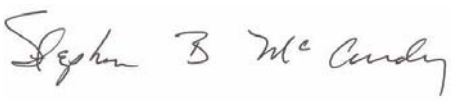
The threat is real, and our instinct is to hide, to focus on the immediate tasks that absorb our time. We try to ignore the demon and hope that it passes over our house. The absence of bad news reassures us that we are safe. After 2006 in which we quietly celebrated that Barth syndrome had not taken a single child, recent months brought the sudden death of Michael Reece in Australia, and serious medical challenges to other families from Australia to Florida, Massachusetts and Utah. Barth syndrome has forced two children to have heart transplants and as I write this, two more are on transplant waiting lists. Others from around the world have been hospitalized with other serious complications of Barth syndrome. Our small community is being starkly reminded that this is still a deadly disorder that threatens each of our affected children and indirectly our extended Barth families.

So where do our families find solace, hope and the courage to carry on?

We find it in you; each and every one of you. We find it in the Reece family who must find it in each other. We find it in the families who post their questions and prayers on our family listserv every day. We find it in the thousands of faithful family, friends and supporters who send their hard earned money to help us in the only way they know how. We find it in the caring eyes of the doctors who struggle to make sense of the complicated aspects of this disease about which still too little is known. And we find it in the passion of the scientists working long hours in laboratories hoping to find the clue that will unlock the puzzle that is still Barth syndrome. We find it most strongly in the determination of leaders like Shelley Bowen who despite her own family's challenges has adopted all of us into her family and refuses to leave any of us behind in the fight for our lives.

The Barth Syndrome Foundation (BSF) is now eight years old and no longer a neophyte as a charity. We have accomplished so much in such a short time. But at our core, all of the people who comprise the Barth syndrome organizations share a simple value. We are a family. We care deeply about each other. We give each other the strength, courage and hope to live, to laugh and to face whatever may come each day, knowing that we are not alone. Our mission is … "*Saving lives through education, advances in treatment and pursuit of a cure for Barth syndrome".* We will never give up.

And I am more proud of that than of any of our many accomplishments (which I have also proudly listed in the following pages!) Thank you. All of you.



Stephen B. McCurdy
Chairman
Barth Syndrome Foundation

Valerie M. Bowen ('Shelley')
President
Barth Syndrome Foundation

BSF 2007 Accomplishments

Family Services *(Led by Shelia Mann and Chris Hope)*

Family Services finds and cares for our families, giving them the strength, knowledge and tools to care for their own. The latest census shows that BSF has grown to a total of 114 boys/young men around the world, with 11 more awaiting a confirmed diagnosis. When new families reach out to BSF, Shelia, Chris and Shelley Bowen are there to welcome and support them. Many of our newer families tell us that they were anxious about contacting us for the first time, not knowing what they would find. Those who do, find a warm voice and a gentle introduction to a new family. In due course, they are introduced to the listserv—our global lifeline on the internet and the place where no question is too small, answers come quickly and everyone understands.

In late 2005, we sought to improve the quality of the listserv by publishing a set of rules governing postings. Usage dropped by more than 50%. After surveying our families we learned that the "rules" made them reluctant to post their questions and thoughts. We immediately rescinded the "rules" and usage of the listserv has nearly returned to its previous levels. We may make mistakes in good faith but we learn quickly!

1n 2007, we experimented with webinars, hosting live, interactive sessions with an expert on a particular subject of interest to the families. The pilot was successful and we are planning more such sessions in 2008.

BSF produces fact sheets, brochures and papers written by investigators who have conducted research to address specific issues within our community such as bullying, depression, hospitalizations, life balance and education, just to mention a few. Last year, the Family Services team created binders containing all of these materials and mailed them to each family. Updated periodically, these binders become an invaluable resource for record keeping, information for new family doctors, contact lists, emergency information for baby sitters and extended family alike. And now, we have upgraded our listservs to be able to send documents electronically to all families or physicians who have signed up.

A visit to an emergency room is always a stressful occasion made even more so if the nurses and doctors have never heard of Barth syndrome. Our latest brochure for healthcare professionals, endorsed by clinical members of our Scientific and Medical Advisory Board and intended for physicians, has been especially well received. It provides a quick and credible source of information that can prove critical in caring for a Barth syndrome patient for the first time. It also serves to encourage the medical staff to consult closely with the parents of the Barth syndrome patient in their care. A copy of this brochure is included in this annual report.

Individuals Associated With The Barth Syndrome Foundation and Affiliates Worldwide										
	Living	Deceased	Pending Diagnosis	Living Individuals By Age in Years						
				0-4	5-9	10-14	15-19	20-29	30-39	40-49
Asia	4	1	2	2	1	0	1	0	0	0
Canada	8	8	1	0	1	3	2	2	0	0
Europe	15	28	2	3	5	1	3	1	1	1
Oceania	8	8	0	0	2	0	4	1	1	0
S. Africa	2	1	0	0	0	2	0	0	0	0
UK	16	7	2	6	5	3	2	0	0	0
USA	61	39	4	11	18	13	9	10	0	0
Total	**114***	**92**	**11**	22	32	22	21	14	2	1
**Living individuals shown by age groups to the right*										

As much as BSF is an internet community, there is nothing like the opportunity to spend time together face-to-face. Our outreach program creates mini-BSF conferences in regions around the country. In August of 2007, the Dunn and Monahan families organized and hosted the BSF Northeastern Outreach event outside of Boston, MA. Over 50 Barth syndrome family members attended along with several physicians and scientists who presented updates on their work and information of use to the families. The Mayor of Brockton declared it *Barth Syndrome Awareness Weekend* and the group spent a terrific weekend attending a Brockton Rox Baseball game on Saturday night and a wonderful family picnic at the Dunn's on Sunday.

BSF Conference (*Led by Jan Kugelmann, Dr. Matt Toth, and Shelley Bowen)*

Planning for the 2008 Scientific, Medical and Family Conference began as the 2006 Conference was coming to a close. Jan Kugelmann led the search, site visits, selection and booking of the location for this year's Conference in Clearwater, Florida, assisted by Leslie Buddemeyer and Shelley Bowen. Jan, Matt and Shelley have been in full planning and organization mode for over a year, mapping the schedules for the individual tracks for scientists and physicians, parents, affected men and boys and their siblings, confirming acceptance by the distinguished speakers, organizing child care and social activities, and the myriad details that make this Conference the consistently professional and successful event that it always is.

Science and Medicine (*Led by Dr. Matt Toth, BSF's Director of Science)*

Research Grant Program

At the beginning of 2006, our Scientific and Medical Advisory Board (SMAB), supported by outside experts, evaluated nine proposals requesting funding for scientific research into Barth syndrome in the 2006 grant cycle. Following the SMAB's recommendations and after thorough review, the BSF Board approved and funded seven grants for a total of $309,200, which are reflected in our 2007 financial statements. These grants have been reported on in our previous newsletter and some of the researchers' results will be discussed at the upcoming Conference in July.

In January of this year, the 2007 grant cycle was completed. Nine highly qualified grant recipients were selected from among the largest number of applications ever received by BSF, and awarded $330,000—the largest commitment of funds for research ever awarded by BSF and its affiliates in a single year. The success of this program, now in its sixth year, bears witness to the growing reputation and impact that BSF and its affiliates are creating in the scientific community. This year's award winners and their research studies are reviewed in an article written by Dr. Toth on pages 7—9 of this annual report. We are excited by the progress that these researchers are making, and the prospects for greater understanding of Barth syndrome improve daily. Equally exciting is that research funded by BSF is having an impact on other disorders and creating linkages that we hope will lead researchers in other disorders to make discoveries that will provide insights back to Barth syndrome as well!

Barth Syndrome Registry and DNA Bank

This program has just completed its second year under the leadership of our co-investigators: Dr. Carolyn Spencer, now at Children's Hospital in Boston, MA and Dr. Barry Byrne at the University of Florida. With the help of the Institute of Child Health Policy at the University of Florida and their expertise in building medical databases, we are now entering the second phase of the project—augmenting the self reported data initially gathered from 43 families, with detailed data abstracted from the patients' medical records. This databank, linked to DNA samples from each child now kept in our biorepository, will prove an invaluable resource for researchers and will provide a much needed baseline with which to evaluate any future therapies. Clearly, more data from more affected individuals and their families increases the impact this effort can have on understanding the evolution of Barth syndrome in patients over time and linking the clinical manifestations to possible genetic variations found in the matching DNA samples. Families attending the clinics held prior to the upcoming BSF Conference in Clearwater, Florida will be able to directly advance the future of scientific research by updating or making their initial contributions to the Barth Syndrome Registry and DNA Bank.

Scientific and Medical Advisory Board

In July of 2007, Dr. Matt Toth convened a full day meeting of scientists and researchers interested in Barth syndrome to map out the most promising scientific paths to accelerate discoveries that will lead to improved treatments and eventually a cure for Barth syndrome. Also in attendance were six members of the BSF Scientific and Medical Advisory Board. The group identified the highest priorities for accelerating research including the need for a mammalian model (e.g. a mouse or a rat with Barth syndrome), specialized antibodies that will help identify the presence of human tafazzin proteins, and high throughput assays that can be used in areas such as newborn screening. This "roadmap" is already guiding BSF investments including grants and contract research into all three areas identified above.

Physician Awareness (*Led by Steve Kugelmann)*

During 2007, BSF and its affiliates sent representatives to and/or set up our booth at eight medical conferences attended by pediatric physicians who may well diagnose and treat a Barth patient. Conferences at which BSF had a visible presence included:

- Child Cardiology 2007 (Children's Hospital of Philadelphia), Orlando, FL
- American College of Medical Genetics, Nashville, TN
- Society for Inherited Metabolic Disorders, Nashville TN
- Pediatric Academic Societies, Toronto Canada (BSF & BSF of Canada)
- United Mitochondrial Disease Foundation, San Diego CA
- Clinical Cardiovascular Genomics, Cold Spring Harbor, NY
- American Heart Association, Orlando FL
- American Society of Hematology, Atlanta GA

Maintaining a presence at these conferences may entail setting up the BSF booth, handing out brochures and answering questions about Barth syndrome and encouraging Barth syndrome researchers to deliver scientific presentations and posters, all designed to increase awareness of Barth syndrome, its symptoms and BSF and its affiliates. Many of our new families are diagnosed or introduced to BSF as a direct result of their physicians learning about Barth syndrome at these meetings. BSF representatives at these conferences included Steve Kugelmann, Jan Kugelmann, Shelley Bowen and Dr. Matt Toth.

Contributions

BSF raised $674,000 from almost 1,000 individual contributors in 2007. These financial supporters are so vital to the continued health of BSF. Without your contributions, none of the important programs described above would be possible. Your donations fund advances in research. You pay for our volunteers to attend the medical conferences. You fund physician awareness and the cost of operating and maintaining our website and listservs—such vital links for our families and physicians. Our upcoming BSF Conference at the Belleview Biltmore Resort in Clearwater, FL will cost over $150,000 to run and is a great investment.

Equally critical are the families and friends who led the fund raising efforts that raised this money. People don't give to institutions or charities, they give to a cause they have come to believe in. A rare and obscure disorder like Barth syndrome is not a high profile cause. There are no celebrities endorsing BSF on Oprah, and most people have never heard of it let alone known someone who is affected. Except our donors.

Every one of them knows someone with Barth syndrome personally or has been told the story by a good friend who does. Every donor was asked to donate to BSF by someone. These are our heroes. The ones who ask. The ones who tell their friends and family of the challenge of Barth syndrome and the hope of BSF. The ones who understand that without financial support, progress stops, families are condemned to isolation and hope dies. So our thanks go to our donors and we ask you to please keep BSF at the forefront of your generosity, even in these difficult economic times. But we reserve our highest praise and deepest appreciation for our fund raisers, for they keep the lights shining… literally!

- Scott Oldewage — Scott's boss, Ed Pace continues to support Scott's colleagues at Lake City Trucks and Lake City International in Utah, who contribute to BSF through their workplace giving program. LCT and LCI match all employee donations and Ed Pace makes a personal donation in each month that his companies are profitable.

- Randy Buddemeyer and the C. B. Richard Ellis Annual Charity Golf Tournament, which once again has set a new record for money raised by CBRE for BSF, doubling their previous year's donation. Randy and Leslie are great spokespersons for BSF, and their friends and business colleagues respond to their requests for donations… and have fun playing golf as well!

- Jan and Steve Kugelmann's annual BSF Drive for a Cure Golf Tournament — This is the granddaddy of BSF golf tournaments as it enters its fifth year in 2008. Jan and Steve have a loyal following who show up every year to have fun, watch the local police helicopter do the Chopper Dropper and to raise money for BSF. It's a labor of love for Jan, Steve and friends.

- For the seventh straight year the McCurdys have sent a simple letter to a growing list of friends from high school, college and graduate schools, work, church and parents of their children's friends from school and around town. The letter makes a simple case, from the McCurdy's hearts asking their friends to once again include BSF in their charitable giving… and every year, their friends respond positively. They say, "We are happy to help in this small way. We know how important this is to you and Will."

- Sue Wilkins started the Woody Varner Fund for BSF in honor of her Dad and added her Mom's name after she passed away in 2007. The Paula and Woody Varner Science & Medicine Fund has been a major fund raiser for BSF and a wonderful way to create a lasting memorial for two people who were so loved by so many.

- Liz and John Higgins love to bowl. So do their friends. Out of such simple pleasures came the annual Bowling for Barth fundraiser which is fast becoming an institution in Highland Lakes, NJ. Who says fundraising has to be hard?

- Tom and Laurie Monahan seem to find an endless number of ways to raise money for BSF, usually involving the Brockton Rox, sports events and the Charlie Horse Sports Bar! No matter what the event, Tom's tireless promotion and great sense of humor and Laurie's organizational skills insure that every event is memorable… and a fundraising success.

- Barth syndrome does not run in Gary Rodbell's family, but Gary doesn't ever seem to stop running for BSF. Running, swimming and biking that is. He is gathering a growing herd of triathletes who train with him and raise money for BSF. In 2007 he and fourteen other athletes raised money for BSF by competing in the Westchester Triathlon in NY. In 2008 some of these iron-people are running in a half Ironman in Laconia, New Hampshire in August and many more are entered again in Westchester, NY on September 21. Coach Gary and "Team Will" guarantee another "win" for Barth!

- When Amer and Jay Randall's triplets had their fourth birthday, the Randalls celebrated the occasion with a party and invited all of their friends to add to the festivities with a gift for the Barth Syndrome Foundation. Since two of the triplets are affected by Barth syndrome, and all of their friends had been asking what they could do to help, this was a natural and welcome way to mark the day.

- Over the last year, in addition to the Paula and Woody Varner Fund, Memorials were established by the families of Tony Satula a friend of the McCurdys in Larchmont, NY and Rob Lochner, Lynda Sedefian's brother in upstate New York. I hope that we can all be as strong and as generous as these families, to think of others despite the loss and sadness they are experiencing. We are eternally grateful.

Research Grants Showcase Increased BTHS Awareness and Interest

The latest cycle of the Barth Syndrome Foundation (BSF) Research Grant Program marks the sixth anniversary of this important funding program with the largest monetary commitment of any previous cycle (over USD $333,000). Six of the nine awardees have not received funding from BSF before.

The 2007 cycle also received the largest number of grant applications which, along with the increasing numbers of publications and of website hits, demonstrates that Barth syndrome (BTHS) research is becoming better known in the scientific and medical community, as well as to the public.

The nine grant recipients for the 2007 cycle are approaching their research from several different angles. Most of the grants are investigating how tafazzin dysfunction causes changes using cellular models in yeast, in cardiac cells, and in white blood cells. Two projects are investigating if there are BTHS patients that have been overlooked, another seeks to make a rat model of BTHS, while another looks into how cardiolipin works on the molecular level.

The following is a summary of the main focus of each individual grant with some thoughts of what value it will bring towards achieving our ultimate goal. The number and breadth of these awards clearly demonstrates the real progress and the increased exposure of BTHS research to the scientific and medical community.

In addition this year‘s cycle has greatly benefited from the increased funding provided by the Barth Syndrome Foundation of Canada and by the Barth Syndrome Trust (UK and Europe). Our worldwide family of organizations continues to grow and mature into something we can all be proud of.

Miriam Greenberg, PhD, Professor and Associate Dean, Wayne State University, Detroit, MI

“Perturbation of the osmotic stress response in cardiolipin-deficient mutants”

Prof. Greenberg uses brewer‘s yeast as a model system to uncover the biochemical connections between the lipid cardiolipin and the genes that affect cardiolipin such as tafazzin—the gene which, when mutated in humans, causes BTHS. Using certain yeast mutants that are deficient in cardiolipin and which have a shortened lifespan, Prof. Greenberg has discovered other mutated genes that are able to restore this lifespan. After testing heat stress, osmotic stress, and screening 80 stress pathway mutants, only mutations in the HOG1 pathway were able to suppress this lifespan shortening of the original cardiolipin-deficient mutants.

HOG1 is a gene found by other investigators and is involved in a cell’s response to stress like the lack of water. HOG1 stands for high-osmolarity glycerol. Because cardiolipin is dysfunctional in BTHS patients, this discovery shows that we may be able to find other pathways and other genes that can ameliorate some of the problems associated with cardiolipin deficiency. How is HOG1 relevant to BTHS? The HOG1 pathway is equivalent to the P38 MAP Kinase in human cells, and we know that P38 MAP Kinase is involved with cardiomyopathy, a defining characteristic of BTHS. These observations point to a parallel equivalence between yeast and humans which is not evident at first impression. Prof. Greenberg seeks to extend and build on these observations by examining the expression and phosphorylation of the HOG1 gene product and its protein targets. Using this information (e.g. inhibition of the P38 MAP Kinase), testable hypotheses for finding treatments for BTHS can be envisioned.

Carol P. Moreno-Quinn, MD, PhD, Assistant Professor, Medical College of Wisconsin, Milwaukee, WI

“Creation of a rat model of Barth syndrome”

The 2006 Nobel Prize in Medicine was awarded to Mello and Fire for their discovery of “RNA Interference.” Using techniques that exploit this Nobel Prize-winning idea, Dr. Moreno-Quinn will make a strain of rat that is deficient in tafazzin expression. Specifically, Dr. Moreno-Quinn will add a transgene to the rat genome that lowers tafazzin expression using RNA interference—a “knockdown” of the tafazzin gene. This relatively new technology is expected to recapitulate the defects that patients with BTHS endure. Dr. Moreno-Quinn will then analyze these knockdown rats to determine if their physiology resembles that experienced by BTHS patients. A mammalian model of BTHS will be a powerful tool in advancing BTHS research, and it has been a goal of many researchers over the years.

Quan He, PhD, Research Scientist, Henry Ford Hospital, Detroit, MI

“Are reactive oxygen species involved in the development of dilated cardiomyopathy in Barth syndrome?”

Using the same Nobel Prize-winning idea of Mello and Fire, Dr. He will lower tafazzin expression in rat neonatal cardiac cells by siRNA oligonucleotides and monitor the effects. Dr. He believes that tafazzin dysfunction causes cardiolipin loss which generates reactive oxygen species (ROS). These ROS cause changes in cardiac cells which eventually lead to apoptosis and dilated cardiomyopathy—a critical problem in BTHS patients. If these experiments are successful then this cardiac-cell model of BTHS will be very valuable for BTHS research and for heart failure research in general.

Matthew R. G. Taylor, MD, PhD, Associate Professor and Director of Adult Clinical Genetics, University of Colorado Health Sciences Center, Denver, CO

“Prevalence of Barth syndrome in adult cardiomyopathies”

By using mRNA hybridization methods, Prof. Taylor will quickly screen for coding changes in tafazzin mRNA in over 200 samples from his Center‘s collection of adult cardiomyopathy patients. If there are positive hits in this registry, DNA sequencing of the tafazzin gene will be used to identify the mutation. If Prof. Taylor finds adults with tafazzin mutations, then conventional ideas about the incidence frequency for BTHS will have to be re-examined. Because BTHS is a very rare genetic disease, reliable frequencies for its incidence are difficult to obtain. There are only a few cases of men with BTHS that are in their 30’s and 40’s. Finding more of these patients will increase our understanding of BTHS and may lead to clues about how to effectively deal with the condition.

David Dale, MD, Professor of Medicine, University of Washington, Seattle, WA

“Neutropenia in Barth syndrome”

Building on previous BSF-funded research, Prof. Dale will use the promyelocytic cell line HL60 to establish a cellular model of neutropenia caused by BTHS. Using a shRNA vector that transiently decreased tafazzin mRNA expression, Prof. Dale will now make a permanent cell line that can be better analyzed for the traits expected in neutropenia. This cell line model of BTHS will approximate the conditions seen in BTHS neutrophils and will be a vehicle to test hypotheses designed to ameliorate this type of neutropenia.

Taco Kuijpers, MD, PhD, Professor, University of Amsterdam, Amsterdam, The Netherlands

“Neutropenia in Barth syndrome: new in vitro models to study BTHS neutrophils”

Using compounds that poison or disable mitochondria function, Prof. Kuijpers will expose the promyelocytic cell line HL60 and will then monitor various parameters that simulate the conditions observed in neutrophils from BTHS patients. Prof. Kuijpers’s hypothesis implicates a defect in electrical potential across the mitochondrial membrane—an observation for which he has provided evidence. It is hoped that these experiments will bring about a more mechanistic understanding of how neutropenia occurs in BTHS.

Richard Epand, PhD, Professor, McMaster University, Hamilton, ON, Canada

“Consequences of the alteration of cardiolipin structure on the properties of the mitochondrial membranes”

With the help of a synthetic chemist, Prof. Epand will examine the titration or hydrogen donor behavior of various cardiolipin species, both symmetric and asymmetric. This biophysical behavior will be analyzed in different membrane curvature environments and in the presence of certain proteins. By understanding what biophysical differences are occurring in membranes with different amounts and different types of cardiolipin, Prof. Epand should be able to show a mechanism by which cardiolipin dysfunction may cause many of the BTHS symptoms.

Susan Kirwin, Senior Research Associate, Assistant Director, Nemours Children‘s Clinic, A. I. duPont Hospital, Wilmington, DE

“Barth syndrome testing: Are we missing some patients?”

By using tafazzin mRNA analysis from blood samples of BTHS and non-BTHS patients, Ms Kirwin will determine if mRNA differences can be used as a diagnostic factor for BTHS. Using data obtained from a clinically diagnosed BTHS patient, Ms Kirwin has shown evidence for an mRNA dysfunction but without a coding mutation in the tafazzin gene. This result means that the spectrum of mutations at the tafazzin gene that gives rise to BTHS has to be expanded to include these types of non-coding mutations. These mRNA results may lead us to derive a correlation between tafazzin mRNA quality and the severity of the symptoms of BTHS patients. A better understanding of what is a normal and what is an abnormal mRNA pattern for tafazzin expression will lead to the identification of patients who have been misdiagnosed in the past, and it may tell us more about tafazzin mRNA expression and BTHS symptoms.

Christopher R. McMaster, PhD, Professor, Dalhousie University, Halifax, Nova Scotia, Canada

“Synthetic genetics towards understanding Barth syndrome cell biology”

Dr. McMaster will use a robotic screening platform to systematically cross the tafazzin deletion strain of yeast with 5,000 other yeast mutants to determine if any of these 5,000 genes interact with tafazzin. By using systematic genetics to identify which genes interact with tafazzin, he will attempt to uncover potential modifiers of tafazzin dysfunction which could lead to new ideas about a treatment for BTHS.

Governance

BSF is governed by our Board of Directors led by our Chairman, Steve McCurdy and our President, Shelley Bowen. Board members serve three-year terms and starting 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:

The Barth Syndrome Foundation Board of Directors and Officers		
Name	**Address**	**Position/Current Term End**
Valerie M. Bowen (Employee)	205 Puckett Road Perry, FL 32348 (850) 584-2712	President and Board Member Term Expiration 2009
Stephen B. McCurdy (Volunteer)	12 Carleon Avenue Larchmont, NY (914) 834-1771	Board Chairman Term Expiration 2010 Chief Financial Officer
Randy Buddemeyer (Volunteer)	4616 W Browning Avenue Tampa, FL 33629 (813) 832-1198	Board Member Term Expiration 2011
Michaela Damin (Volunteer)	1 The Vikings Romsey, Hampshire S051 5RG 44 179 4518 785	Board Member Term Expiration 2010
Stephen Kugelmann (Volunteer)	7145 Briar Oak Drive Merritt Island, FL 32953 (321) 453-4585	Board Member Term Expiration 2010
Katherine R. McCurdy (Volunteer)	12 Carleon Avenue Larchmont, NY (914) 834-1771	Board Member Term Expiration 2011
Susan S. Osnos (Volunteer)	272 Round Hill Road Greenwich, CT 06830 (203) 622-0472	Board Member Term Expiration 2009
Susan V. Wilkins (Volunteer)	6219 Barbara Lane Lincoln, NE 68512 (402) 421-1926	Board Member Term Expiration 2011
Lynda M. Sedefian (Employee)	101 Stone Ridge Court Altamont, NY 12009 (518) 452-6939	Secretary (Officer)
Matthew J. Toth, PhD (Employee)	132 Creemer Avenue Iselin, NJ 08830 (732) 283-3417	Director, Science & Medicine

Scientific and Medical Advisory Board

In addition, BSF benefits greatly from the advice and counsel of our Scientific and Medical Advisory Board (SMAB), a world class group of distinguished scientists and physicians with expertise in the diverse fields of research and the multiple systems affected by Barth syndrome. These advisors review all research grant proposals and make their recommendations to the BSF Board who retain final grant approval authority. In addition, they advise on scientific and medical programs including awareness and BSF's semi-annual International Conference, and offer clinical consultation to our families' doctors when asked.

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	**Iris L. Gonzalez, PhD** Molecular Diagnostics Lab *(retired)* A. I. DuPont Hospital for Children Wilmington, Delaware	**Jeffrey A. Towbin, MD** Pediatric Cardiology Texas Children's Hospital Baylor College of Medicine Houston, Texas
Peter G. Barth, MD, PhD *Emeritus* Pediatric Neurology Emma Children's Hospital / AMC *(retired)* Amsterdam, The Netherlands	**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
Barry J. Byrne, MD, PhD Pediatric Cardiology Shands Children's Hospital University of Florida Gainesville, Florida	**Grant M. Hatch, PhD** Lipid Lipoprotein Research University of Manitoba Winnipeg, Canada	**Gerald F. Cox, MD, PhD** Clinical Genetics Children's Hospital Boston, Massachusetts Senior Medical Director, Clinical Research, Genzyme Corp. Cambridge, Massachusetts
Michael Schlame, MD Anesthesiology NYU School of Medicine New York, New York	**Salvatore DiMauro, MD** Neurology Columbia University College of Physicians and Surgeons New York, New York	**Colin G. Steward, FRCP, FRCPCH, PhD** Pediatric Hematology Bristol Royal Hospital for Children Bristol, England
Katherine R. McCurdy *ex officio* Board Member Barth Syndrome Foundation Larchmont, New York	**Matthew J. Toth, PhD** *ex officio* Science Director Barth Syndrome Foundation Iselin, New Jersey	

Barth Syndrome Registry and DNA Bank

The Barth Syndrome Registry and DNA Bank is operated under contract for BSF by the University of Florida and led by Dr. Carolyn Spencer MD and Dr. Barry Byrne. BSF and Dr. Spencer have established a separate advisory board for the Barth Syndrome Registry and DNA Bank to insure free and open access as well as compliance with the requirements of the University of Florida's Institutional Review Board (IRB).

Barth Syndrome Registry and DNA Bank Advisory Board	
Carolyn Spencer, MD Co-Principal Investigator Department of Cardiology Children's Hospital Boston, Massachusetts	**Barry J. Byrne, MD, PhD** Co-Principal Investigator Pediatric Cardiology Shands Children's Hospital University of Florida Gainesville, Florida
Richard I. Kelley, MD, PhD Division of Metabolism Kennedy Krieger Institute Johns Hopkins University Baltimore, Maryland	**Melissa Maisenbacher, MS, CGC** Medical Data Abstraction, *ex-officio* University of Florida College of Medicine Gainesville, Florida
Michael Schlame, MD Department of Anesthesiology New York University School of Medicine New York, New York	**Deepa Ranka, MS** IT and Statistical Research Analyst, *ex-officio* Department of Epidemiology and Health Policy Research University of Florida College of Medicine Gainesville, Florida
Colin G. Steward, FRCP, FRCPCH, PhD Pediatric Hematology Bristol Royal Hospital for Children Bristol, England	**Gerald Cox, MD, PhD** Clinical Genetics Children's Hospital Boston, Massachusetts Senior Medical Director, Genzyme Corporation Cambridge, Massachusetts
Matthew J. Toth, PhD Barth Syndrome Foundation Science Director Iselin, New Jersey	

International Affiliates

The Barth Syndrome Foundation has several international affiliates which together comprise the International Barth Syndrome Foundations, a group of independently incorporated entities bound by a common vision, shared and closely coordinated programs and the authorized use of the BSF logo, written materials, internet websites and other materials. These affiliates are:

The 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	

The Barth Syndrome Foundation is a member of the Genetic Alliance. Approval by the Better Business Bureau is also in progress.

The Barth Syndrome Foundation is a member of the the National Health Council, improving the health of all people, particularly those with chronic diseases and/or disabilities.

Note: Complete copies of BSF's annual reports, audited financial statements, Form 990s and Board minutes are available on BSF's website at www.barthsyndrome.org.

SCHALL & ASHENFARB Certified Public Accountants, LLC

IRA L. SCHALL, CPA
DAVID C. ASHENFARB, CPA

INDEPENDENT AUDITORS' REPORT

To the Board of Directors of
The Barth Syndrome Foundation, Inc.

We have audited the accompanying statement of financial position of The Barth Syndrome Foundation, Inc. (the "Foundation") as of December 31, 2007 and 2006 and the related statements of activities, functional expenses, and cash flows 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, based on our audits, 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, 2007 and 2006, and the changes in its net assets and its cash flows for the years then ended in conformity with accounting principles generally accepted in the United States of America.

Schall & Ashenfarb

Schall & Ashenfarb
Certified Public Accountants, LLC

March 7, 2008

350 Fifth Avenue, Suite 728
New York, New York 10118
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THE BARTH SYNDROME FOUNDATION, INC.
STATEMENT OF FINANCIAL POSITION
AT DECEMBER 31, 2007 AND 2006

	12/31/07	12/31/06
Assets		
Cash and cash equivalents (Notes 2g and 2h)	$2,317,189	$2,111,135
Investments (Note 5)	286,833	11,044
Accounts receivable	1,477	548
Unconditional promises to give (Note 2c and 2e)	973	336,305
Prepaid expenses	7,297	1,299
Total assets	$2,613,769	$2,460,331
Liabilities and Net Assets		
Liabilities:		
Accounts payable and accrued expenses	$15,684	$12,153
Grants payable	168,300	119,024
Total liabilities	183,984	131,177
Net Assets: (Note 2b)		
Unrestricted	1,993,659	1,939,939
Temporarily restricted	436,126	389,215
Total net assets	2,429,785	2,329,154
Total liabilities and net assets	$2,613,769	$2,460,331

The attached notes and auditors' report
are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
STATEMENT OF ACTIVITIES
FOR THE YEARS ENDED DECEMBER 31, 2007 AND 2006

	12/31/07			12/31/06		
	Unrestricted	Temporarily Restricted	Total	Unrestricted	Temporarily Restricted	Total
Public Support and Revenue:						
Contributions (Notes 2c and 3)	$291,849	$382,608	$674,457	$656,463	$362,425	$1,018,888
Interest income	116,505		116,505	66,816		66,816
Net assets released from restrictions:						
Satisfaction of program restrictions (Note 3)	335,697	(335,697)	0			0
Total public support and revenue	744,051	46,911	790,962	723,279	362,425	1,085,704
Expenses:						
Program services	596,095		596,095	550,838		550,838
Management and general	81,864		81,864	98,875		98,875
Fundraising	12,372		12,372	4,115		4,115
Total expenses	690,331	0	690,331	653,828	0	653,828
Change in net assets	53,720	46,911	100,631	69,451	362,425	431,876
Net assets - beginning of year	1,939,939	389,215	2,329,154	1,870,488	26,790	1,897,278
Net assets - end of year	$1,993,659	$436,126	$2,429,785	$1,939,939	$389,215	$2,329,154

The attached notes and auditors' report
are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
STATEMENT OF FUNCTIONAL EXPENSES
FOR THE YEAR ENDED DECEMBER 31, 2007

	Program Services	Management and General	Fundraising	Total 12/31/07	Total 12/31/06
Salaries	$191,188	$25,561		$216,749	$109,779
Payroll taxes and benefits	13,869	1,854		15,723	9,413
Total personnel services	205,057	27,415	0	232,472	119,192
Research grants	309,200			309,200	163,801
Professional	20,774	27,842		48,616	124,283
Telephone	2,453	3,055		5,508	4,304
Office expense	7,691	9,451	8,766	25,908	46,549
Printing and publications	8,271	2,683	1,526	12,480	9,477
Dues and conferences	4,241	4,518		8,759	18,574
Transportation	26,036	4,536		30,572	84,320
Insurance		2,043		2,043	2,298
Meals	5,987	321	2,080	8,388	53,892
Audio visual expense				0	22,461
Exhibits	6,385			6,385	4,677
Total expenses	$596,095	$81,864	$12,372	$690,331	$653,828

The attached notes and auditors' report
are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
STATEMENT OF FUNCTIONAL EXPENSES
FOR THE YEAR ENDED DECEMBER 31, 2006

	Program Services	Management and General	Fundraising	Total 12/31/06
Salaries	$95,584	$11,356	$2,839	$109,779
Payroll taxes and benefits	8,073	1,072	268	9,413
Total personnel services	103,657	12,428	3,107	119,192
Research grants	163,801			163,801
Professional	88,443	35,840		124,283
Telephone	2,240	1,929	135	4,304
Office expense	29,254	16,825	470	46,549
Printing and publications	5,354	4,123		9,477
Dues and conferences	6,950	11,584	40	18,574
Transportation	71,553	12,404	363	84,320
Insurance		2,298		2,298
Meals	52,448	1,444		53,892
Audio visual expense	22,461			22,461
Exhibits	4,677			4,677
Total expenses	$550,838	$98,875	$4,115	$653,828

The attached notes and auditors' report
are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
STATEMENT OF CASH FLOWS
FOR THE YEARS ENDED DECEMBER 31, 2007 AND 2006

	12/31/07	12/31/06
Cash Flows from Operating Activities:		
Change in net assets	$100,631	$431,876
Adjustments to reconcile change in net assets to net cash provided by/(used for) operating activities:		
Donated stock	(10,395)	(21,573)
(Increase)/decrease in assets:		
Accounts receivable	(929)	0
Unconditional promises to give	335,332	(36,305)
Prepaid expenses	(5,998)	1,035
Increase/(decrease) in liabilities:		
Accounts payable and accrued expenses	3,531	(10,234)
Grants payable	49,276	(15,271)
Total adjustments	370,817	(82,348)
Net cash provided by operating activities	471,448	349,528
Cash Flows from Investing Activities:		
Proceeds from sales of investments and donated stock	21,596	41,148
Purchase of investments	(286,990)	0
Net cash (used for)/provided by investing activities	(265,394)	41,148
Net increase in cash and cash equivalents	206,054	390,676
Cash and cash equivalents - beginning of year	2,111,135	1,720,459
Cash and cash equivalents - end of year	$2,317,189	$2,111,135

Supplemental data:
Interest paid - $0
Income taxes paid - $0

The attached notes and auditors' report are an integral part of these financial statements.

THE BARTH SYNDROME FOUNDATION, INC.
NOTES TO FINANCIAL STATEMENTS
DECEMBER 31, 2007 AND 2006

Note 1. 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 act as a public foundation, operated for the following purposes: a) to promote awareness of Barth Syndrome; b) to educate and support physicians, research centers and organizations addressing the causes, diagnosis, treatment and cure of Barth Syndrome, and c) to assist in the support of families with children suffering from Barth Syndrome.

The Foundation is a tax-exempt organization under Section 501(c)(3) of the Internal Revenue Code. They have not been designated as a private foundation.

Note 2. Summary of Significant Accounting Policies

a. Basis of Accounting

The financial statements have been prepared on the accrual basis of accounting and accordingly reflect all significant receivables, payables, and other liabilities.

b. Basis of Presentation

The Foundation follows Statement of Financial Accounting Standards (SFAS) No.'s 116 and 117, *Accounting for Contributions Received and Contributions Made*, and *Financial Statements of Not-for-Profit Organizations*. Under SFAS No. 117, organizations are required to report information regarding their financial position and activities according to the following classes of net assets:

- *Unrestricted* – represents all activity without donor imposed restrictions as well as activity with donor imposed restrictions, which expire within the same period.

- *Temporarily restricted* – relates to contributions of cash and other assets with donor stipulations that make clear the assets restriction, 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, 2007 and 2006.

c. 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.

d. Donated Assets

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

e. Unconditional Promises to Give

Unconditional promises to give are recognized as revenues or gains in the period received and as assets or decreases of liabilities, depending on the form of the benefits received. Conditional promises to give are recognized when the conditions on which they depend are substantially met. At year end, the full amount is due within one year. These receivables have been evaluated for collectability and no allowance for doubtful accounts is deemed necessary.

f. In-Kind Contributions

Under SFAS No. 116, organizations are required to recognize contributions of services that enhance non-financial assets and require specialized skills. Many individuals volunteer their time and perform a variety of tasks that assist the Association with specific programs, campaign solicitation, and various committee assignments. These volunteer services do not meet the criteria stated above and have not been recognized in the financial statements.

g. Cash and Cash Equivalents

For purposes of financial reporting, cash and cash equivalents include cash held in banks, certificates of deposits and money market funds. There were no interest or taxes paid during the year.

h. Concentration of Credit Risk

The Foundation maintains its cash balances at institutions they consider to be credit worthy. The cash balances are insured by the Federal Deposit Insurance Corporation (FDIC) for amounts up to $100,000. At December 31, 2007 and 2006, the Foundation had uninsured cash balances of $0 and $26,713, respectively.

i. Capitalization Policies

Items of property and equipment with an individual cost in excess of $5,000 are recorded 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.

j. Expense Allocation

The costs of providing various programs and other activities have been summarized on a functional basis in the statement of activities and in the statement 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 other specific function but provide for the overall support and direction of the Foundation.

k. Management Estimates

The preparation of financial statements in accordance 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. Temporarily Restricted Net assets

At December 31, 2007, temporarily restricted net assets consist of the following:

	Balance 1/1/07	Contributions	Released from Restrictions	Balance 12/31/07
Program Restrictions:				
Paula & Woody Varner Science and Medicine Fund	$89,215	$99,703	$0	$188,918
Barth Syndrome Trust	0	55,000	(35,697)	19,303
Science and Medicine Fund	0	227,905	0	227,905
Total Program Restrictions	89,215	382,608	(35,697)	436,126
Time Restrictions	300,000	0	(300,000)	0
Total	$389,215	$382,608	($335,697)	$436,126

At December 31, 2006, temporarily restricted net assets consist of the following:

	Balance 1/1/06	Contributions	Released from Restrictions	Balance 12/31/06
Paula & Woody Varner Science and Medicine Fund	$26,790	$62,425	$0	$89,215
Time Restrictions	0	300,000	0	300,000
Total	$26,790	$362,425	$0	$389,215

Note 4. Commitments

Grants payable accrued of $168,300 are all due to be paid within twelve months.

Although the Foundation does not have any long-term leases, they are committed to research grants awarded subsequent to year end, up to the amount of $333,454, that are payable in 2008 and 2009.

Note 5. Investments

Securities are carried at fair market value in accordance with SFAS No. 124. At December 31, 2006, investments consisted of shares in Exxon Mobil Corp. and American Express Company. At December 31, 2007, investments consisted of a United States Treasury Bill. At year end, the quoted market price was used as the basis of determining fair value, which also approximates the original cost.

www.barthsyndrome.org