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## Birth of New BSF Research Era

*Please meet two of the many 'faces' of Barth syndrome, who happen to be brothers!*



**By Kate McCurdy, VP Science and Medicine**

The Barth Syndrome Foundation, Inc. has now completed the review of another highly successful round of research grant applications, and I am extremely pleased to highlight those that have been awarded. Once again, BSF received excellent applications from investigators around the world. The BSF Board of Directors voted to accept five of these this year and to award research funds totaling \$163,801. The details of these research grants can be found on *page 6* of this issue.

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## A Conference Agenda to Stimulate Barth Syndrome Research and Understanding

**By Kate McCurdy, VP, Science & Medicine**

Dr. Richard I. Kelley, M.D., Ph.D., leading the team preparing the agenda for the Scientific and Medical meetings of the July 2006 International Barth Syndrome Conference, writes, 'The conference will bring together most of the world's experts on Barth syndrome and join them for two days with other leaders in specific aspects of

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Science and Medicine  
Barth Syndrome Foundation, Inc.  
New York

# Transforming Speculation to Science

By Valerie "Shelley" Bowen, President

Dear Friends,

This newsletter marks a period of momentous milestones, not only for the Barth Syndrome Foundation, but also for Barth syndrome. It was in April ten years ago when the gene for Barth syndrome was first described in a journal article by Bione et al. This discovery came twenty-three years after Barth et al first described the disorder. The discovery of this gene was identified within the first year of the multi-centered effort to map the human genome. With the discovery of the gene, a tool was delivered to transform speculation into a diagnosis. It seems poignant that we close the first of our five-year strategic plans by unveiling the final accomplishments of the major goals achieved within this plan at this 10-year anniversary date of the discovery of the gene *G4.5*.

As we embark on a renewed strategic plan to plot our priorities for the next five years, I think it appropriate to reflect upon another article published in 1996 by Sokal. Around the same time period when Bione's investigations were quietly published in *Nature Genetics*, this other article gained considerable attention in the journal *Social Text*, a journal primarily focused on the social sciences. The *Social Text* had eliminated the peer review process for selected publications in their journal. The reasoning behind this was to promote a less conventional approach to report research. The journal trusted authors of prospective articles to guarantee the academic integrity of their work.

The notoriety of this article was not based upon the merit of the science but on the lack thereof. On the date of the article's release, Dr. David Sokal, a physicist at New York University, announced to the world that his article was a hoax. This scandalous act, now referenced as '*The Sokal Affair*,' has been hailed by some as brilliant and conversely as fraudulent and unethical. In 1998, Dr. Sokal retrospectively wrote about this event. He asserted, "*If one does not take into account empirical aspects, the scientific discourse indeed becomes nothing more than a myth or narration.*"

Regardless of one's opinion of Dr. Sokal's methods, he certainly managed to make his position clear. There is a significant distinction between science and speculation. The parallel of Sokal and Bione is an interesting one. Like Bione, Dr. Sokal was most concerned about the importance of standards in methodology of scientific research as well as the process which safeguards the integrity of articles regarded as science. Both are committed to publish science opposed to speculation. The scientists in our community well-know the difference between science and speculation. We have relied heavily upon the credibility of our SMAB to meticulously scrutinize research proposals on behalf of those we love for the past four years. Scientific progress about Barth syndrome is dependent upon the integrity of scientific investigation and discovery. As a lay group it would be very easy for our organization to be caught off-guard by scientific proposals that lack scientific merit.

It was a volunteer mother, Kate McCurdy, who maintained a keen eye to detail in creating a scientific research plan. She included safeguards to protect BSF with appropriate measures of peer review to advise us on the merit of science versus unwittingly awarding a grant because we were desperate for progress. It is important to realize how this process and our advisors have protected our interests over the years.

(Cont'd on page 3)

(Cont'd from page 2)

Not only have these selfless men and women overseen a program which offers great promise for our loved ones, they have also played a vital role in our education about the significance of research with an approach of repetitive methodical rigor. However, make no mistake there is merit for speculation in the genesis of science. The collective wisdom of families through the exchange of information on our listserv has enlightened the scientific community about trends never before reported on in this disorder. It is through this exchange, in some cases, that hypothesis driven research has been launched and has in fact, enlightened us and the medical community about life-threatening risks associated with Barth syndrome.

In this newsletter you will learn about the establishment of the Barth Syndrome Medical Database and Biorepository. This approach, as Michaela Damin simply states, "...will transform the stories into science," and provide an opportunity for families to proactively take part in moving beyond speculation to science. Investigators will launch research efforts through this collaboration and significantly benefit with the ready access to the data and biospecimens required to carry out their studies.

You will also learn of our most recent grants awarded supporting sound research. These arduous efforts of unlikely partners has led to a highly-regarded research grant program and has led to a significant increase in journal articles relating to Barth syndrome in peer-reviewed, well-known scientific journals. We have also outlined our enormous strides in seeking the funds required to carry out our ambitious scientific and medical strategic plan, and our success thus far in securing these funds.

While the 'Sokal Affair' targeted a community investigating "soft science," his assertions, regardless of political position, are astute. Throughout the historical timeline of Barth syndrome and BSF, investigators and our stakeholders have invested in ideas. The return on their investments in time, talent and funds has been ideas realized. I would like to thank every person who has ever invested resources to assure the success of our organization's five-year plan. It has been one wild ride. Five years ago, the five founders of BSF dreamed of a better future for those with Barth syndrome. Today, because of methodical research, allocations of appropriate resources and a well-organized five-year plan for BSF, those dreams have become a reality. Are you ready to make dreams come true? If so, jump in and take part in what promises to be another remarkable five-year investment into the future.



BSF is a proud member  
of the National Health Council



For all correspondence, including information, please contact:

**BOARD OF DIRECTORS:**

**VALERIE (SHELLEY) BOWEN**

PRESIDENT  
sbowen@barthsyndrome.org

**STEPHEN KUGELMANN**

VICE PRESIDENT, AWARENESS  
skugelmann@barthsyndrome.org

**KATHERINE R. MCCURDY**

VICE PRESIDENT, SCIENCE AND MEDICINE  
kmccurdy@barthsyndrome.org

**STEPHEN B. MCCURDY**

CHAIRMAN AND CHIEF FINANCIAL OFFICER  
smccurdy@barthsyndrome.org

**SUSAN V. WILKINS**

BOARD MEMBER AT LARGE  
swilkins@barthsyndrome.org

**ADDITIONAL CONTACTS:**

**LYNDA M. SEDEFIAN**

EXECUTIVE ADMINISTRATIVE ASSISTANT  
NEWSLETTER EDITOR  
lsedefian@barthsyndrome.org

**LEONARD STEINBERG, EA, CMC**

BOOKKEEPER

**MICHAEL HOPE**

WEBMASTER  
mhope@barthsyndrome.org

**SHELIA MANN, smann@barthsyndrome.org**

**CHRIS HOPE, chope@barthsyndrome.org**  
CO-CHAIRS, FAMILY SERVICES

**BSF AFFILIATES**

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

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

Barth Trust of South Africa  
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

BSF's newsletter is designed for educational purposes only and is not intended to serve as medical advice. The information provided within this newsletter should not be used for diagnosing or treating a health problem or disease. It is not a substitute for professional care. If you suspect you or your children may have Barth syndrome you should consult your health care provider.

All submissions and correspondence regarding the newsletter should be directed to Lynda Sedefian at: lsedefian@barthsyndrome.org.

# A Summary of BSF's Progress in 2005

By Valerie "Shelley" Bowen, Executive Director

## 1) To insure that all appropriate medical professionals are aware of Barth syndrome and have ready access to the latest tools to make a timely and accurate diagnosis.

- a) Journal Articles
  - i) Four journal articles published that acknowledge BSF for grant support  
(1) Gonzalez<sup>1</sup>, Valianpour<sup>2</sup>, Xu<sup>3</sup>, Schlame<sup>4</sup>
  - ii) Three additional articles published that mention BSF and/or BSF families involvement  
(1) Spencer<sup>5</sup>, Bateman, Huhta<sup>6</sup>
- b) Outreach Program
  - i) Birmingham Children's Hospital, UK
  - ii) Great Ormond Street Hospital, UK
  - iii) Newcastle upon Tyne Children's Hospital, UK
  - iv) Emory Medical Center, Department of Medical Genetics, Atlanta, GA USA
  - v) Emma Children's Hospital, Amsterdam NL
  - vi) Necker Children's Hospital, Paris FR
  - vii) Primary Children's Hospital, Salt Lake City, UT USA
- c) Professional Conferences
  - i) 8th Annual Update on Pediatric Cardiovascular Disease, Orlando, FL USA
  - ii) 2005 American College of Human Genetics Annual Clinical Genetics Meeting, Dallas, TX USA
  - iii) Pediatric Academic Societies' Annual Meeting, Washington DC, USA
  - iv) 16th Annual American Society of Echocardiography Scientific Sessions, Boston, MA USA
  - v) American Heart Scientific Sessions, Dallas, TX USA
  - vi) 4th Annual American Society of Hematology Meeting, Atlanta, GA USA
- d) Media
  - i) Television Feature Documentary; National  
(1) Discovery Health Channel, Mystery Diagnosis, "Blood Brothers"
  - ii) Television News Feature Story; Regional  
(1) Barth Syndrome Foundation Outreach, WPVI; Philadelphia, PA USA  
(2) Barth Syndrome Foundation, Live at Five, WCTV; Tallahassee, FL USA

- iii) Print News; Regional
  - (1) Bingo Fundraiser to Aid BSF, TACO Times; Perry, FL USA
  - (2) Funds for BSF, Madison Enterprise-Recorder; Madison, FL USA
  - (3) Taylor County Fun Day to Aid BSF, TACO Times; Perry, FL USA
- iv) Print Periodical Publication; South Eastern US
  - (1) Sportsman Unite for a Cure, Woods-N-Water Magazine; Perry, FL USA

## 2) To stimulate the development of successful treatments for Barth syndrome (a multi-system disorder) and enable their delivery.

## 3) To encourage, guide and fund additional research to improve diagnosis and treatment, and ultimately to develop a cure for Barth syndrome.

- a) Awarded 5 research grants totaling \$163,801
- ## 4) To create a caring and informed community of Barth families actively involved in supporting each other and our organization.

- a) Created multiple professionally reviewed Fact sheets on various components of BTHS to educate families about the complexities of this disorder
- b) Formalized and expanded BSF Membership, to be used as a tool to determine needs of families:  
2005 Year-End Worldwide Membership Statistics  
Living (99)  
Deceased (72)  
Deaths in 2005 (1)  
Awaiting Confirmed Diagnosis (26)  
Diagnosed in 2005 (8)  
New to BSF (21) 27% increase from 2004

## 5) To build and sustain a broad base of committed contributors who will provide the funds we need to achieve our vision.

- a) (See Fundraising for BSF, page 16)

## 6) To inspire and make effective use of an organization dedicated to reaching our vision.

- a) Capacity Building
  - i) Hosted 5th BSF Volunteer Enrichment Workshop
- b) Membership with Umbrella Groups
  - i) National Health Council  
(1) Received certificate of membership

(Cont'd on page 5)

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- from the National Health Council in meeting all 41 standards of excellence
- ii) Genetic Alliance
    - (1) First member organization to be featured in the Genetic Alliance "Member Success Spotlight" for model organizations
    - (2) First member group to participate in "Incubator Program"
  - c) International Affiliates
    - i) Canada receives charitable status becoming the Barth Syndrome Foundation of Canada
    - ii) South Africa receives charitable status becoming the Barth Trust of South Africa
    - iii) BST UK entering third year as a charity
  - d) National Institutes of Health
    - i) Participated in and led a portion of the 2005 National Heart, Lung and Blood Institute (NHLBI) Public Interest Organization (PIO) annual meeting
    - ii) Participated/presented at NHLBI/ORD Working Group on Cardiomyopathies of Rare Diseases
    - iii) Member of CETT Program Review Board
    - iv) Meetings held with representatives of ORD, NHLBI (Heart Division), NHLBI (Blood Division), NIAMS, NINDS
  - e) Outreach
    - i) Salt Lake City, UT USA
    - ii) Clearwater, FL USA
    - iii) Philadelphia, PA USA
    - iv) Newcastle, Bristol, Romsey, Birmingham, London, UK
    - v) Amsterdam, NL
    - vi) Paris, FR

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- (1) Gonzalez IL. **Barth syndrome: TAZ gene mutations, mRNAs, and evolution.** Am J Med Genet A 2005 May 1;134(4):409-14.
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- (4) Schlame M, Ren M, Xu Y, Greenberg ML, Haller I. **Molecular symmetry in mitochondrial cardiolipins.** Chem Phys Lipids 2005 December;138(1-2):38-49.
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# Birth of New BSF Research Era

(Cont'd from Cover)

## **Principal Investigator:**

**Miriam L. Greenberg, Ph.D.**  
**Professor, Biological Sciences**  
**Associate Dean for Research**  
**Wayne State University**  
**Detroit, MI**

## **Project:**

***Does Copper Deficiency Play a Role in Barth Syndrome?***  
**(\$40,000 awarded over 1 yr.)**

“Barth syndrome (BTHS) is a severe X-linked disorder characterized by cardiomyopathy, skeletal myopathy, neutropenia, growth retardation, and abnormal mitochondria. The defining cellular features of BTHS are mutation in the tafazzin gene and aberrant cardiolipin (CL) composition. However, substantial phenotypic variation occurs even among patients with the same tafazzin mutation, indicating that the clinical outcome is strongly influenced by other physiological factors. The current gap in our knowledge of the identity of these modifying factors complicates the diagnosis and treatment of BTHS.

This study will test the hypothesis that perturbation of CL synthesis leads to copper deficiency and increased oxidative damage. This hypothesis is based on preliminary findings that a yeast mutant lacking CL exhibits copper deficiency. We will measure copper levels and markers of oxidative damage in CL mutants and determine if copper alleviates mutant defects.

Copper deficiency has been shown to cause cardiomyopathy and neutropenia in humans and animals. Experimental support for the proposed hypothesis would warrant follow-up studies of cellular

and mitochondrial copper in BTHS patients. A finding of copper deficiency in BTHS would have important implications for the treatment of this disorder.”

## **Principal Investigator:**

**Thomas H. Haines, Ph.D.**  
**Professor, Chemistry and**  
**Biochemistry**  
**City College of the City University of New York**  
**New York, NY**

## **Project:**

***A Role for Cardiolipin in Barth Syndrome***  
**(\$40,000 awarded over 1 yr.)**

“Barth syndrome appears to be due to a defect in cardiolipin metabolism. The established symptoms may be due to a deficiency in ATP in selected tissues, especially heart and skeletal muscles, retarded growth and neutropenia. The result of insufficient ATP synthesis in these selected tissues during development and in response to activities of those afflicted with the defect would specifically produce such symptoms. The purpose of this study is to provide definitive chemical evidence for a proposed conformation of the headgroup of cardiolipin that implies a role in ATP synthesis. A defect in cardiolipin structure that is critical to the role it plays in ATP synthesis may be important when approaching a cure.

We have developed a model for the role of cardiolipin in ATP synthesis that is due to a unique conformation of the lipid in bilayers. The conformation is distinct from that in solution or in any mixture that is not a bilayer or membrane. There are

six mitochondrial proteins involved in ATP production, all of which bind cardiolipin. Our conformation of the headgroup suggests that a critical role of the lipid is to provide protons that are pumped by the three-mitochondrial proton pumps and also to provide the protons to the consumers of the gradient (F<sub>0</sub>F<sub>1</sub> ATPase, the phosphate carrier and the ATP/ADP exchange protein). Although the present evidence strongly supports the notion that this conformation is correct, definitive proof is necessary before taking the insight to the clinical level. Although it is widely believed that cardiolipin is associated with mitochondrial ATP synthesis, we do not know how. Our model suggests how.

In order to unequivocally prove our conformation of cardiolipin in bilayers, we will synthesize a cardiolipin molecule with 18O in its phosphates. This will permit us to compare the details of the IR to that of the natural 16O cardiolipin. The comparison of the spectra will permit us to show that the conformation in the bilayer state is distinctly different from that of the cardiolipin under any other circumstances and that the predicted conformation is that consistent with the model. The second set of experiments, which will be conducted simultaneously with the IR experiments, is to use NMR to seek two protons that hold our model together. This NMR approach has only recently been made available for seeking high-energy protons in proteins. We will apply it to cardiolipin. Measurements must be made on bilayers. Thus we require solid state NMR.

This work will further our knowledge about Barth syndrome.”

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**Principal Investigator:**  
**Grant M. Hatch, Ph.D.**  
**Professor, Pharmacology and**  
**Therapeutics**  
**Acting Associate Dean of**  
**Medicine for Research**  
**University of Manitoba**  
**Winnipeg, Manitoba, Canada**

**Project: *Cholesterol Metabolism***  
***in Barth Syndrome***  
**(\$40,000 awarded over 2 yrs.)**

“Barth syndrome is a rare X-linked genetic disorder in young boys. The disease is caused by mutations in the tafazzin (*TAZ*) gene and mutations in *TAZ* result in alterations in cellular cardiolipin metabolism and a moderate hypocholesterolemia among other phenotypic abnormalities. The reason for the hypocholesterolemia observed in Barth syndrome is unknown. Hypocholesterolemia has been observed in some genetic disorders and evidence suggests that cholesterol metabolism may be altered in these. Given that cardiolipin may play a role in cellular cholesterol homeostasis and that cholesterol itself plays a key role in embryogenesis, alterations in functional *TAZ* expression may affect cholesterol biosynthesis and this may have serious consequences for organ and tissue development in Barth syndrome boys. It is thus important to understand how cholesterol metabolism is altered in Barth syndrome. With this knowledge, we may begin to not only understand how alterations in *TAZ* may affect metabolic enzyme systems but how these alterations may be reversed. In this project, we seek to determine how altered *TAZ* expression regulates cholesterol metabolism.

We hypothesize that reduction in the level of functional *TAZ* and that mutations in *TAZ* lead to a reduction in cholesterol de novo biosynthesis in Barth syndrome cells and this is mediated by a reduction in the expression of transcription factors and cholesterol biosynthetic enzymes. Moreover, we believe that addition of functional *TAZ* to Barth syndrome cells, which may have a reduced cholesterol metabolism, will restore cholesterol metabolism to that of normal cells.”

**Principal Investigator:**  
**Toshihide Kobayashi, Ph.D.**  
**Chief Scientist**  
**Director of Lipid Biology**  
**Laboratory**  
**RIKEN**  
**Wako, Saitama, Japan**

**Project: *Characterization of***  
***Endosome Specific Lipid***  
***Lysobisphosphatidic Acid in***  
***Lymphoblasts from Patients***  
***with Barth Syndrome***  
**(\$23,100 awarded over 1 yr.)**

“Barth syndrome is an X-linked disease caused by mutations in tafazzin, a putative phospholipid acyltransferase. The disorder causes a primary defect in remodeling of cardiolipin (CL) and its precursor, phosphatidylglycerol (PG). Whereas metabolism of PG to CL is well established, our recent results indicate that PG is also a precursor of late endosome/lysosome specific lipid, lysobisphosphatidic acid (LBPA, also called as bis(monoacylglycero)phosphate, BMP). LBPA plays important roles in organization of late endosomes, lysosomal lipid degradation and transport of lipids and proteins from late endosomes. At present, little

is known about LBPA metabolism and function of endosomes in cells from Barth syndrome. In this study, we will examine the metabolism of LBPA, the ultrastructure of late endosomes/lysosomes, the endocytic function and the cholesterol homeostasis of lymphoblasts from patients with Barth syndrome.”

**Principal Investigator:**  
**Eric A. Storch, Ph.D.**  
**Assistant Professor, Psychiatry**  
**and Pediatrics**  
**University of Florida**  
**Gainesville, FL**

**Project: *Psychosocial Aspects***  
***of Barth Syndrome: Quality of***  
***Life, Family Functioning, and***  
***Relationship to Biologic***  
***Variables***  
**(\$20,701 awarded over 1.5 yrs.)**

“Barth syndrome, also known as x-linked cardiomyopathy and neutropenia, is a rare but serious genetic disorder that affects males. Although the past two decades have seen an increase in research on the genetic and biological aspects of the disease, only one preliminary report has examined psychosocial functioning in boys with Barth syndrome. Anecdotal accounts from clinicians and parents of boys with Barth syndrome have suggested that these families face an array of stressors related to the chronic illness that impacts a variety of realms (e.g., sibling and peer relations, school performance, mood and behavior, parental stress). This study will expand upon such reports and our knowledge about the impact of other chronic pediatric illnesses to examine the

(Cont'd on page 8)

# Birth of New BSF Research Era

(Cont'd from page 7)



## BSF Research Grant Program

Please note that the details of the 2006 Barth Syndrome Foundation, Inc. Research Grant Program will be available on our website at: [www.barthsyndrome.org](http://www.barthsyndrome.org) later this spring.

The deadline for submission of applications will be sometime in the fall.

specific psychosocial factors that affect risk and resilience in boys with Barth syndrome and their families. A variety of psychosocial variables will be assessed in an attempt to capture the unique experiences of families with a son who has Barth syndrome. Our findings will have implications in understanding psychosocial variables that may impact quality of life and treatment adherence, as well as identifying areas in which psychosocial intervention may be beneficial.”



With these awards, BSF now has supported a total of nineteen research projects, in five countries, for a total financial commitment of over \$640,000, and this has all occurred since 2002 when our grant program began. At least nine journal articles are in print already as the direct result of BSF-funded research.

Our already productive research grant program will become even more powerful when it is coupled with the IRB-approved Barth Syndrome Medical Database and Biorepository that BSF has just signed a contract to create (*see article about these on page 12*). The fact that BSF soon will be able to offer a triad of resources – funding, biological samples and linked clinical data will make researching Barth syndrome even more desirable.

Once again, I want to take this opportunity to thank everyone who has made this possible. That includes the scientists and physicians who conduct the research, the experts who review the applications, the BSF Board of Directors who make the tough decisions with the best interests of all those affected by Barth syndrome in mind, those who work hard to raise the money so that we can have such an effective program, and those of you who have generously donated the funds required to make it all possible. Our continued team effort is our best hope for a bright future. I am counting on its success.



## BSF Science Director Job Search

As many of you know, the BSF Board of Directors has decided to make a significant investment in our future and to seek an individual with a Ph.D. to become our Science Director. This person will administer the BSF research grant program, coordinate the scientific portions of the international Barth syndrome conferences organized by BSF (like the one that will take place this July) and develop and coordinate the various operating and strategic scientific efforts of the Foundation. Volunteers at BSF have done a terrific job advancing our cause in these areas to date, but we all agree that it is time to hire a professional to spearhead these initiatives. I am extremely excited about this big step in our evolution and very much appreciate the Board's acceptance of my request for the creation of this new position.

The search is going very well. At this writing, we have received applications from 37 candidates, many of whom are excellent. We have reviewed their written materials and now are conducting a series of interviews. If all goes according to plan, I eagerly look forward to introducing this new member of our "Barth family" at the conference.

Kate McCurdy, VP Science and Medicine

# A Conference Agenda to Stimulate Barth Syndrome Research and Understanding

(Cont'd from Cover)



Kate McCurdy,  
Vice President,  
Science and  
Medicine; and ex  
officio member of  
BSF's Scientific  
Medical Advisory  
Board

science and medicine most relevant to the understanding and treatment of Barth syndrome. We anticipate that the many formal presentations and other opportunities for discussion of recent advances and discoveries in Barth syndrome will generate new research ideas that would not otherwise have developed. The results of the meeting should help focus the investigators on the most important problems to be solved and thereby chart much of the next several years of research on Barth syndrome<sup>1</sup>.

The conference will include formal presentations and multi-disciplinary forums for discussion, brainstorming and collaboration. The first day, July 6, will follow a scientific theme. Miriam L. Greenberg, Ph.D. and Michael Schlame, M.D. will lead the morning session entitled "**Gene Products and Biochemistry of Barth Syndrome**." This will include talks by Mindong Ren, Ph.D., Arnold W. Strauss, M.D., Peter Rehling, Ph.D., Eric Testet, Ph.D. and Tal M. Lewin, Ph.D., as well as the Chairs. The afternoon meetings will be led by Richard I. Kelley, M.D., Ph.D. and will address "**New Frontiers of Barth Syndrome Science**." Francois P. Bernier, M.D., Robert E. Jensen, Ph.D. and Grant M. Hatch, Ph.D., will also deliver presentations.

The second day will concentrate more on the complex clinical aspects of the disorder. The morning will bring a series of talks on the "**Cardiac Issues of Barth Syndrome**", led by Jeffrey A. Towbin, M.D. Carolyn T. Spencer, M.D., Arnold W. Strauss, M.D., Randall M. Bryant, M.D., Robert E. Shaddy, M.D. and Tal Geva, M.D. will also speak. The following session, chaired by Dr. Steward, will focus on the "**Hematological Aspects of Barth Syndrome**" and include presentations by Colin G. Steward, M.A. (Cantab) B.M., B.Ch. (Oxon), F.R.C.P., F.R.C.P.C.H., Ph.D., Taco W. Kuijpers, M.D. and David C. Dale, M.D.

The third day will comprise working sessions to develop further treatment guidelines for Barth syndrome. By making these guidelines accessible to physicians around the world it is hoped that clinical outcomes will improve and lives will be saved. There will also be discussions about the newly created Barth Syndrome Medical Database and Biorepository.

We are looking forward to welcoming you to this stimulating and productive conference. For registration details see [www.barthsyndrome.org](http://www.barthsyndrome.org).

*Kate McCurdy, VP Science and Medicine*



## 2006 Family Conference: Inspirational, Educational, and Supportive

*By Jan Kugelmann, Chair, Conference Committee*

Inspirational, educational and supportive – our 2006 family agenda vows to be all these, and more. As promised, we have planned a top-notch educational symposium on Barth syndrome, and as in the past we will provide unique one-on-one opportunities for families to interact with experts in multiple subspecialties.

Day one of the family meetings will focus on cardiology and the newly formed Barth Syndrome Medical Database and Biorepository (see page 12), as well as a "Jeans to Genes Overview" moderated by Iris Gonzalez, Ph.D., a true champion behind Barth syndrome – both in science and friendship.

During day two, attendees will get to meet many of the researchers funded by BSF and discuss the complexities of, and new developments in, their research efforts. We will also delve into the hematological and neurological aspects of Barth syndrome, with addresses by Colin Steward, Ph.D., and Ariel Sherbany, M.D., to name a few.

(Cont'd on page 10)

# 2006 Family Conference...

(Cont'd from page 9)

***"I have never attended a conference before where I immediately bonded with other families and shared my hopes and fears for my son so easily.***

***The physicians and scientists who have attended the conferences are the most dedicated group of medical personnel I have met and I now have resources available that I never dreamed I would find."***

***~ Susan Hone, Mother***

On day three, we plan to roll up our sleeves and address the psychosocial issues of the day-to-day aspects of a chronically ill child. Families will also have the unequalled opportunity to talk with educators, psychologists and pediatricians about their concerns during our "break-away" sessions.

## **Back by Popular Demand!**

The break-away sessions are a wonderful opportunity for parents to meet in small groups with others whose children are the same age and are dealing with similar issues. Topics to be addressed range from early intervention to transitioning into adult healthcare; from entering kindergarten to preparing for college; the challenge of getting healthy foods into your toddler, to accepting the fact that your son is now a young man and ready to make his own decisions.

And there's more! I am pleased to announce two very exciting

additions to our 2006 conference agenda which are sure to leave lasting impressions. **First:** Renowned pediatric cardiologist Dr. Roberto Canessa from Montevideo, Uruguay will be our keynote speaker on Thursday afternoon. In October 1972, Roberto Canessa was with his rugby team aboard an ill-fated flight from Argentina to Chile that crashed in the Andes Mountains. He and a companion reached civilization more than two months later, and their survival and rescue were later featured in a well-known book and film entitled "Alive". He has recently been highlighted on the NBC's *Today Show* and in the April issue of *National Geographic Adventure* magazine. Hear his amazing story of courage and survival – *Human Groups in Crisis Situations*. **Next:** He has played Carnegie Hall in New York City and the Sydney Opera House in Australia. From church basements to fire halls, synagogues to Las Vegas showrooms, Taylor Mason uses stand-up comedy and his own brand of ventriloquism to make every audience, every age, laugh. He has headlined all the major comedy clubs in the United States and is featured on the Disney Cruise Line as well as hosting his own children's television show "*Taylor's Attic*". Comedian, musician, ventriloquist, entertainer and actor, Taylor will join us on Friday evening for our international themed social event!

It's not too late to register! Please make your hotel reservations via our website at [www.barthsyndrome.org](http://www.barthsyndrome.org) where you will find more detailed information about Barth syndrome and this year's conference. If you have any questions or need further

information, please contact myself, Jan Kugelmann, via email at: [conference2006@barthsyndrome.org](mailto:conference2006@barthsyndrome.org)

We look forward to seeing you in Orlando this summer - The City Beautiful! You are guaranteed memories that will last a lifetime!



**What our previous attendees are saying:**

***"I was totally impressed. This was the best-organized conference I have ever been to. As a grandparent, I felt the conference was really informative and helpful ... a wonderful experience. I'm looking forward to the next one!"***  
***Jerre Vogt, Grandmother***



*They came together, formed friendships, and made their statement - we will persevere!*

***"The conference experience is unparalleled. When families meet, it's as though parents, children and sibs already have a unique "shorthand" which validates the daily struggles and triumphs of those affected by Barth Syndrome."***

***Maria Olson, Aunt***

# BSF Financial Health Sparks Optimism for the Future

By Steve McCurdy, Chairman and CFO



2005 marked the fifth full year of life for the Barth Syndrome Foundation, and our success to date gives us a stronger sense of optimism for the future than ever before. For BSF to have a meaningful impact on the lives of boys, young men, and families affected by Barth syndrome we had to build a strong community

for our families, become a credible advocate and leader in the worlds of medical science and treatment, assemble and nurture a dedicated group of volunteers, and convince donors to care enough to support all of our dreams with their money. By virtually every measure, our Foundation continues to make great progress on every one of our key goals.

Financially, we finished the last year stronger than ever before, and are well positioned to increase our investments in a number of important long-term Science and Medicine programs. We raised over \$1.4 Million and ended the year with net assets of \$1.9 Million (an increase of almost \$1.1 Million over 2004). BSF awarded \$157,000 in research grants in 2005 and invested over \$276,000 in program services. The higher figure for programs in 2004 includes a \$130,000 investment in our BSF Conference, held once every two years. This Conference will be held again in July 2006. Fundraising and administrative costs represented 27.6% of our total expenses, as we created the infrastructure for several new programs in 2006. Non-program expenses will absorb a much smaller share in 2006. A summary of our financial statements, audited in 2004 and 2005 by Schall & Ashenfarb, Certified Public Accountants LLC., can be seen on page 12.

We have recently been accepted as a member of the National Health Council, having met its 41 standards of excellence in governance, and stand in the company of much larger and more established foundations such as the American Heart Association, the American Cancer Society, and the American Diabetes Association. We now register every year with some 14

states in the US. Taken together with the 12 states that require no registration, BSF is now free to conduct fundraising campaigns in 26 states. We will shortly pass a very important milestone as we have successfully raised a sufficient percentage of our donations from public sources over the last five-year trial period to earn a permanent designation as a 501 (c) (3) charitable organization by the U.S. Internal Revenue Service. Each of these accomplishments represent an important recognition of our credibility as a foundation that is here to stay and making steady progress toward our goals.

You may recall that at the end of last year we launched a multi-year Science and Medicine fundraising campaign to supplement our general fundraising efforts. The purpose of this campaign is to try to pre-fund the majority of our expected spending on Science and Medicine programs for the next decade. These programs include our Research Grant Program, our newly created Barth Syndrome Medical Database and Biorepository, our International BSF Scientific/Medical & Family Conference, a series of proposed scientific and medical conferences, and our ongoing physician awareness program.

Thanks to the extraordinary generosity of a number of major donors, we received donations and/or commitments of almost \$1.3 Million in the first few months of the campaign! This is an exceptional vote of confidence in the future of BSF and a tribute to the success of our programs, the dedication and professionalism of our volunteers, and the growth and continuing support of our family community around the world. Make no mistake, we still have a long way to go before we achieve our vision of "...a world in which no one need suffer or perish from Barth syndrome." In our case, passion, caring, dedication and professionalism has proven to be a successful combination of traits, and our success is accelerating!

Our program expenses are budgeted to more than double in 2006 – an investment we would be concerned to make without the benefit of our \$1.8 Million fund balance and the continued support of our broad base of donors. Our ten-year Science and Medical funding needs are estimated at over \$6 Million, so we still have

*(Cont'd on page 12)*



### **Details of how the Barth Syndrome Medical Database and Biorepository will work**

BSF will share the expenses with UF and join in the success of this venture. The Principal Investigator for this project will be Dr. Carolyn Spencer, who will chair the Database and Biorepository. Many of the parents know Dr. Spencer well as she began gathering data from a number of affected individuals at our last BSF Conference in 2004 under a research grant from BSF. The University of Florida Institute of Child Health Policy has extensive experience in the creation, housing and analysis of large databases of health data. The abstracted data

will be entered by a geneticist and data requests will be mined by a biostatistician. An epidemiologist will aid construction of questionnaires to eliminate potential bias in reporting data and work with the biostatistician. These efforts will be overseen by Dr. Elizabeth Shenkman, working with Dr. Spencer.

BSF has agreed with UF that the operation of the Medical Database and Biorepository will be overseen by Dr. Spencer, who will be advised by a group of experts including the head of BSF's Scientific and Medical Advisory Board – Dr. Richard I. Kelley – and a representative of BSF – Shelley Bowen. In addition, in order to insure that all patients and families who will contribute data and samples are fully informed and give their consent to the ownership and use of these samples and data in appropriate research... and that the patients remain anonymous, Dr. Spencer must receive the approval of UF's Institutional Review Board (IRB) before she collects anything. All approvals are expected to be received in time for the BSF Conference in Florida this July, where samples and medical histories will be collected from all attending and consenting families. In time, the rest of the Barth families will also be contacted to invite them to add to this critical resource.

Our initial goal is to collect family history, patient information, test results and DNA specimens, and we may well be able to extend this to collect lymphoblast cell lines obtained through a single blood draw which

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***"From here on,  
you the families affected by  
Barth syndrome, can make a  
real difference to research  
and, the search for treatments  
and a cure.  
Your determination  
to contribute your personal  
histories and samples  
will insure that scientists  
will continue to be drawn  
to Barth syndrome  
and to BSF."***

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would immortalize cell lines for research for years to come. There will be a redundant off-site storage for DNA, lymphoblasts and data. The UF IRB has reviewed our grant process and has agreed to fast-track IRB approval for requests of data and biospecimens received by investigators who have received research funding from BSF.

The addition of the Barth Syndrome Medical Database and Biorepository to the resources we now make available to researchers is a major accomplishment for BSF. Soon, in addition to providing research grants and access to the only comprehensive library of peer-reviewed articles about or related

to Barth syndrome, BSF can now provide the world's largest central database of medical histories of individuals with Barth syndrome and centralized collection of DNA and tissue samples. Funding, medical data, DNA and tissue samples, and the BSF library will be a very tempting collection of resources that BSF believes will attract additional researchers to the study of Barth syndrome, its causes, its treatment, and its cure.

BSF was expertly advised in its effort by John Walsh, CEO of Alpha One, who developed the first agreement of this sort with UF a number of years ago, and by Dr. Jon Merz, PhD, MBA and JD from the University of Pennsylvania. Dr. Merz's fields of specialty include intellectual property, medical ethics and Institutional Review Boards, and he will continue to be an advisor to BSF to insure that our families' interests are fully protected. Also advising BSF was Christine Ethridge from the law firm of Kirkpatrick & Lockhart, Nicholson, Graham, LLC, in Pittsburgh.

From here on, you the families affected by Barth syndrome, can make a real difference to research and, the search for treatments and a cure. Even the longest journey begins with the first step. Will you make the next step by working with us and through your personal histories and samples held in the database assist scientists to study Barth syndrome. To participate please contact Shelley Bowen at  sbowen@barthsyndrome.org.

## NIH Research Initiatives Seeking Applications

In addition to the vast investigator-initiated research that is supported by the National Institutes of Health (NIH), research in some specific areas is solicited by various NIH institutes from time to time. Applications for these usually are accepted for February 1, June 1 and October 1 deadlines every year. ***The following ongoing NIH initiatives are particularly relevant to Barth syndrome.***

### Exploratory and Developmental Research Grants for Investigations in Rare Diseases (R21) (Initiative number: PA-03-171)

**Purpose:** To encourage exploratory and developmental research projects by providing support for the early and conceptual stages of projects that represent novel approaches to the understanding, treating, and preventing rare diseases in the areas of heart, lung, and blood disease, as well as sleep disorders. **Please visit:** <http://grants1.nih.gov/grants/guide/pa-files/PA-03-171.html> for more details.

### Chronic Illness Self-Management in Children (Initiative number: PA-03-159)

**Purpose:** To solicit research related to improve self-management and quality of life in children and adolescents with chronic diseases. Children with a chronic illness and their families have a long-term responsibility for maintaining and promoting health and preventing complications of the chronic disease. Research related to sociocultural, environmental, and behavioral mechanisms as well as biological/technical factors that contribute to successful and ongoing self-management of particular chronic diseases in children is encouraged. **Please visit:** <http://grants2.nih.gov/grants/guide/pa-files/PA-03-159.html> for more details.

### Tools for Zebrafish Research (Initiative number: PAR-05-080)

**Purpose:** To encourage investigator-initiated applications designed to exploit the power of the zebrafish as a vertebrate model for biomedical and behavior research. **Please visit:** <http://grants.nih.gov/grants/guide/pa-files/PAR-05-080.html> for more details.

### Chronic Fatigue Pathophysiology and Treatment (Initiative number: PA-05-030)

**Purpose:** To support investigator-initiated research on the epidemiology, diagnosis, pathophysiology, and treatment of chronic fatigue syndrome (CFS) in diverse groups and across the life span. **Please visit:** <http://grants.nih.gov/grants/guide/pa-files/PA-05-030.html> for more details.



*Kai, age 4, from Bristol, England*



*Bridger, age 6, from Provo, Utah*



*Ben, age 9, from Knoxville, Tennessee, loves his new set of wheels!*

## 'Faces' of Barth syndrome

## What is Barth Syndrome?

Barth syndrome is a rare but serious X-linked recessive disorder, in which the clinical effects of the G4.5 (or TAZ1) gene mutation are manifested only in males. The characteristics of Barth syndrome include the following in varying degrees, even within the same family:

**Cardiomyopathy:** Heart muscle weakness. This, combined with a weakened ability of the white blood cells to fight infections, represents the greatest threat to those individuals with Barth syndrome.

**Neutropenia:** Reduction in the number of "neutrophils," a type of white blood cell that is extremely important in fighting bacterial infections. The neutropenia may or may not follow a regular cycle, but in either case, it puts Barth individuals at an increased risk of serious infections.

**Muscle Weakness and General Fatigue:** All muscles in a Barth individual, including the heart, have a cellular deficiency which limits their ability to produce energy, causing extreme fatigue during activities requiring strength or stamina – from walking, to writing, to growing.

**Growth Delay:** Most boys with Barth syndrome are below average in weight and height, often substantially so, until the late teenage years.

**Early and accurate diagnosis is key to survival for Barth syndrome boys.** Historically, boys died of heart failure or infection by 3 years of age, but today, with improved diagnosis, treatment, and management, the survival rate and future of these boys is much brighter.



## Calendar of Events

### April 2006

- 21-23 BSF Annual Board Meeting
- 22 BStrust Outreach for Dutch-Speaking Families
- 23 BSFCanada Annual General Meeting
- 23 BSFCanada Outreach

### May 2006

- 8 Barth Trust South Africa Presentation at the Reach for a Dream Foundation
- 15 BSFCanada Executive Committee Meeting
- 18 BSF Executive Committee Meeting

### June 2006

- 3 BST Trustees Meeting
- 20 BSF Executive Committee Meeting
- 22 BSFCanada Executive Committee Meeting

### July 2006

- 3-8 BSF Int'l Scientific/Medical & Family Conference; Lake Buena Vista, FL
- 18 BSF Board Meeting

### September 2006

- 11 BSFCanada 2nd Annual Golf Tournament
- 19 BSF Executive Committee Meeting

### October 2006

- 17 BSF Board Meeting
- 18-21 BSF booth @ 35th Annual Child Neurology Society Meeting; Pittsburgh, PA

### November 2006

- 21 BSF Executive Committee Meeting

### December 2006

- 9-12 BSF booth @ The American Society of Hematology 48th Annual Meeting & Expo; Orlando, FL

## Community Fundraisers

### "Birth of Barth" Month

The month of May has been designated as "Birth of Barth" month in honor of our boys and their mothers. Don't forget to collect your loose change for BSF, and set up canisters in your community!

### "Friends of Barth Quilt" Raffle

Raffle tickets are being sold for a Queen/Full Size Quilt, handcrafted by Nina Rivers and given to BSF as a gift to help raise funds. Drawing to be held @ BSF's 2006 International Conference. Prices: 1/\$2; 6/\$10; 10/\$15; 15/\$20; 20/\$25 (Books of 10). Please contact Joyce Lochner ([JOYLOC105@nycap.rr.com](mailto:JOYLOC105@nycap.rr.com)) or Lynda Sedefian ([Lsedefian@barthsyndrome.org](mailto:Lsedefian@barthsyndrome.org)).

### Florida Ironman ~ Panama City, Florida November 4, 2006

Gary Rodbell, John Steigerwald, Matthew Karp and Laurie Parkinson will be punishing their bodies for 140 miles for our Barth Families.

### Longstock Water Garden Open Day August 20, 2006

(near Stockbridge, Hampshire, UK)  
(By kind permission of the John Lewis Partnership)



A warm thank you to Terry Dunbar (father) & Gary Dunbar (son) of Zakspin Spin-casters for the production of BSF's lapel pins. (Terry is "Gramps Terry" to Nick of Romsey, Hampshire); Paolo Ruzzier who did all the laser cutting; and Nori Damin (Nick's grandmother or 'Nonna'), who paid for all the materials. We had a fun day putting them all together when we were out in South Africa on holiday.

# **\$1.444 Million Raised for Barth Syndrome in 2005**

*By Steve McCurdy, Chairman & CFO*

**W**hat a year! The fundraisers for BSF were working overtime in 2005. They raised over \$1.4 Million for BSF and its affiliates' programs in support of affected families, clinicians, researchers, and to keep our Foundation running. They had more success than we have ever had in the past with contributions coming from donors large and small, and from events ranging from sales of Barth Blue Lemonade in the heat of summer, to baked goods, hot chocolate, chocolate chip cookies, and poinsettias during the winter holidays. Events included the Kugelmann's annual BSF Golf Tournament in Merritt Island Florida, the BSF Canada Golf Tournament in Ontario, Canada, the Monahan's increasingly popular Sports Auction in Brockton, Massachusetts, and the Higgins' Bowling Party in Warwick, New York, a few of which are mentioned below.

Eliza McCurdy, the 11 year old sister of Will McCurdy, in a repeat of her 2004 holiday hot chocolate and chocolate chip cookie fundraiser, raised the bar still further in 2005. She and five of her friends from Greenwich Country Day School received permission from Saks Fifth Avenue in Greenwich to conduct her holiday fund raiser in the store's front entrance on a heavy pre-Christmas shopping day. In four hours, Eliza and her friends raised just over \$1,000 from the pleasantly surprised patrons of Saks.

## **The Science and Medicine Fund Off to a Fast Start**

Following the announcement of the kick-off of the BSF Science and Medicine Fund late last year, we have already received over 28 contributions totaling almost \$1.3 Million. Our goal is to raise between \$5-7M to pay for the next 10 years of our research grant program, our international scientific/medical and family conferences, a series of focused scientific conclaves, our physician awareness program, and our new Barth Syndrome Medical Database and Biorepository. We owe a huge debt of thanks to five anonymous donors who showed their faith in BSF's efforts by contributing a total of \$1.2 Million to get us off to a very fast start! Thank you!

## **Growing the Woody Varner Fund**

Woody and Paula Varner are the parents and grandparents of Sue and John Wilkins, respectively.

The former President of the University of Nebraska, Woody passed away in 1999, but Paula has been a friend of BSF from the very start. It was Paula who contributed the money for the first gathering of Barth families in Baltimore in 2000 and who made the very first founding donation to BSF. Stepping up to help again, this time in Woody's memory, Paula and Sue have established the *Woody Varner Science and Medicine Fund* for BSF and have approached many of their friends for support. In a very short time, Paula and Sue have raised \$57,180 from over 70 friends who admired Woody and what he stood for, and have the same respect for Paula and the Wilkins family.

## **Upcoming Fund Raisers in 2006**

### **"Birth of Barth" Month**

May has been designated as "Birth of Barth" month in honor of the boys, their mothers, and in a season of hope and renewal. Families across the world are placing small canisters to collect loose change for BSF in their homes, their offices, and local businesses.

### **Local Community Supports Birth of Barth**

For only the second year, St. John's Church in Larchmont, New York has decided to ask its parishioners to contribute as one to a charity that is close to and important to the community. The Vestry chose BSF since the McCurdy family are a part of the St. John's community, and will be making an Easter Appeal to its parishioners during Pentecost – a period on the Church's calendar that corresponds to "Birth of Barth" month – to contribute to BSF. Proceeds will be contributed to BSF's Science and Medicine Fund.

### **2006 Ironman**

Our incredible Ironmen are at it again! Gary Rodbell and John Steigerwald, this time joined by Matthew Karp and Laurie Parkinson (who are entering their first Ironman!), will be punishing their bodies for 140 miles on Nov. 4 for our Barth Families. They will be returning to the Florida Ironman in Panama City, Florida, seeking to win the Janus Fund Award for the most money raised for a charity by a competitor. This will be the third time that Gary and John have raced for BSF. The first time was in 2002 in Panama City where they raised \$79,000 and came in third among fund raising competitors. The second time was in 2004 in Madison, Wisconsin where

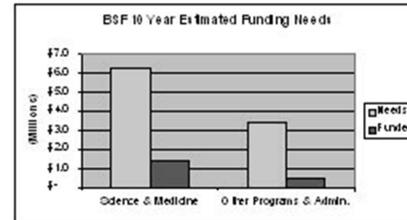
*(Cont'd on page 17)*

(Cont'd from page 16)

they raised almost \$150,000 and came in second among fundraisers. For Gary who is 53, this will be his last Ironman - John who is a still-young 45 will continue until his body crashes! We want to make sure that Gary goes out as the winner we know him to be. We ask all of our friends to make their donations from now through October in support of the 2006 BSF Ironmen. Our goal is to raise over \$300,000 to make our Ironmen winners!

We at BSF are deeply grateful for all of the efforts put forth on our behalf by all of our fundraisers. We are happy to put endless volunteer hours into BSF and affiliate programs, but most of what we accomplish would not ever be possible without the continuing faith

and support of the people whose names appear inside the back cover of this Newsletter. You are the wind beneath our wings!



### BSF Still Needs Our Help!!

We have made a good start in funding our long-term requirements for Science and Medicine, but finding

the funds needed to run all of our programs and keep BSF operating is still a challenge. **We need everyone's help!**



*The Barth Syndrome Foundation, Inc. (BSF) appreciates your contribution. Your gift helps us continue our programs designed to increase awareness; support and educate families and physicians; and fund research. Please visit our website at [www.barthsyndrome.org](http://www.barthsyndrome.org) for more information. All gifts are tax-deductible to the fullest extent permitted by the law. The official registration and financial information of BSF may be obtained from the Pennsylvania Dept. of State by calling toll-free, within PA, 1-800-732-0999. Registration does not imply endorsement. BSF's Florida registration number is SC-12347. One hundred percent of your contribution will be received by BSF. Please forward all contributions to: The Barth Syndrome Foundation, Inc., P.O. Box 618, Larchmont, New York 10538*

## Our Opportunity to Help Other Families

By Shelia Mann and Chris Hope, Co-Chairs, Family Services

Just imagine being told that your child has a potentially fatal disorder and there is no one else to speak to or there is no research being done to find a cure. This happened to some of our very own



Shelia Mann, Co-Chair, Family Services, together with her daughter English (left) and son Ben (right). Shelia and her family are from Tennessee.

Barth families not too long ago. That is why BSF was created, to serve all of those who are affected by this disorder. It is the desire of BSF and the Family Services Team to have no family ever feel that sense of isolation again.

Looking back over the last year, we are encouraged by the number of families that have contacted us and have come to join BSF as members. Our Awareness

program and our website are obviously doing a good job. More and more families are contacting us right after a diagnosis, and often before the results of tests are known. Parents are being proactive in finding solutions for their children, and spreading the message. One of the most important jobs within Family Services is to encourage the contact between families, not only to help out and give support during the struggles which we all face, but also, to hear the good news, so that we can all celebrate together.

We strongly believe that informing affected families through our listserv and Peer Support Program, about the advancements in research, treatments and critical new data has been crucial to the survival of our children. With advances in science, medicine and research the resources and educational tools required to care for our children will become more complex. The BSF Family Service Team will continue to review and update the educational materials and resources that are

(Cont'd on page 18)

# Our Opportunity to Help Other Families

(Cont'd from page 17)



Chris Hope, Co-Chair, Family Services, together with her son Robert, from Ontario, Canada.

offered to affected families and to create additional fact sheets related to Barth syndrome.

We have heard lately of the accomplishments of those with Barth syndrome. They are receiving awards, becoming ambassadors, and in general making themselves heard. In talking to families we have heard that their Barth child is healthier, and has missed less school than their non-affected siblings. The more our boys do, the more they are able to encourage and help each other, and the less Barth syndrome makes them 'different.' As we plan the youth agendas for the conference, we are constantly being reminded that they do not want lessons on Barth syndrome, but they want to play and hang out. This is exactly what any 'normal' child would want to do during their summer vacation.

Chris and I are both parents of a son with Barth syndrome and know all too well how demanding caring for a chronically ill child can be, but volunteering our time is crucial to the well-being of all who are affected. Because of dedicated volunteers like you, our efforts are paying off. We are now seeing our boys turning

into men and facing positive challenges like entering college, independent living and even marriage. They are overcoming challenges never before dreamed possible!

With increased awareness among the medical world our family membership numbers continue to grow, and with this comes the need for more and more volunteers to assist us. Being a volunteer for BSF is very rewarding. Having the opportunity to meet and speak with newly diagnosed families and providing them with the advocacy tools and educational materials needed to best care for their Barth loved one is truly a pleasure. By working together we can move BSF closer to reaching its vision, "A world in which no one will suffer or perish from Barth syndrome". Why don't you join the many others and become a volunteer!

Although Family Services' primary focus is on the lives and doings of the Barth individual, and working

toward improving the quality of their lives, we are also here to provide services for their siblings, and we enjoy hearing about them. When you read the Sibling Spotlight (page 25), you will see that they have full lives. They dance, play sports, take music lessons, and so on. They appreciate and share concerns regarding their brothers, and help out by holding their own fundraisers.

As mentioned, this year of course is a conference year, and we can't stress enough how important this event is for the entire family. We would like to encourage everyone's attendance. Barth clinics, the knowledge and understanding gained from the Family Sessions, and the connection made with other families truly make it the most important service that BSF provides for its families and members.

We look forward to meeting new families and renewing old friendships at the conference this year!



## Discovery Health ~ Mystery Diagnosis...

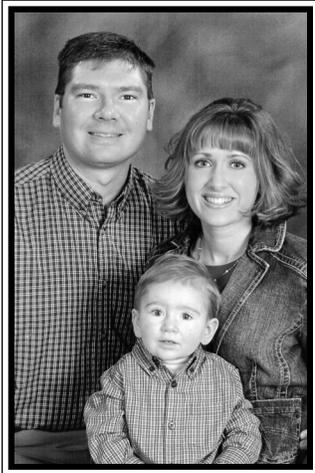


Discovery Health continues to air a special called "Blood Brothers," the Bowen family's struggle to understand the mystery disease that was threatening both of their young sons. Barth syndrome is an under-diagnosed and potentially fatal disorder, and the national viewing of this episode has significantly increased awareness of Barth syn-

drome. BSF would like to thank the Bowen family for volunteering to be a focus of this nationally aired episode. Discovery Health Channel's Mystery Diagnosis website can be found at: <http://health.discovery.com>.

# Focus on New Family

By Tracy Brody, Iowa (Parent of Barth child)



We are the Brody Family from Arcadia, Iowa. My husband, D.W. and I were extremely excited to welcome our son, Bly into our lives on February 16, 2004. The pregnancy and delivery went well, and Bly seemed to be a healthy little boy. He nursed well and gained weight normally; however, we were very concerned about his lack of

sleep and discomfort. At 1.5 months his eating became poor and he began vomiting often. Our doctor said it was acid reflux and/or colic. We were then referred to a pediatric specialist for a second opinion, and an upper GI exam revealed that Bly's heart was enlarged.

We were immediately transferred to a cardiologist in Des Moines, IA. Bly's ejection fraction (EF) was at 8.5%, his heart was taking up 70% of his chest cavity, and he had severe mitral valve leakage. He was put on several heart medications and we were told if his heart didn't shrink within one week he'd need a transplant. Since there was no improvement with Bly's heart after five months, our cardiologist felt a transplant would give him a better chance of making it to his 2nd or 3rd birthday. After discussing Bly's situation with his colleagues, however, it was decided to push his medications harder by nearly doubling all of the amounts. Bly's heart improved significantly at 13 months of age. We were ecstatic!

We met with a GI doctor and an occupational therapist at Mayo Clinic to help with Bly's eating problems. He wouldn't try any baby food or table food and would take his bottle 10-12 times a day, only taking 1-2 ounces at a time. His eating somewhat improved after his reflux issues were being treated. We also had a physical therapist working with Bly. He didn't scoot, crawl or roll over until he was 15 months. He walked on his own at 17 months.

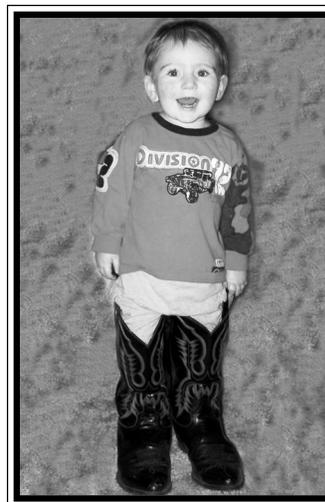
At 19 months, Bly started showing low neutrophil counts. The cardiologist mentioned in his notes that we needed to look into Barth syndrome at our next visit. D.W. and I didn't know what to think. I immediately got online to

find out what Barth syndrome was. I was overwhelmed and confused. Although all of the symptoms fit Bly perfectly, I thought there must be some kind of mistake. No other family member is known to have this disorder.

A few months later we met with a genetic specialist to have testing done. The results came back positive. Bly was diagnosed with Barth syndrome at 23 months. We are so thankful to have a diagnosis. We can meet with the appropriate doctors and we somewhat know what Bly's future holds.

Since Bly's diagnosis, BSF has been amazing with providing us information and support. We've learned a great deal about this rare disorder from BSF's website, newsletters, and the involvement on their listserv. I'm so grateful for the many phone calls and e-mails. The support from BSF is very uplifting and heartfelt.

We are looking forward to attending the 2006 BSF Conference this summer. We are excited to meet other Barth boys and gain a better understanding of Barth syndrome from a medical standpoint. We are also anxious to hear more about the research that is being done and any new advances that have been made. We'd love to help in the advancement of finding a cure for all of our boys.



*"Bly is now 2 years old and is doing well. His heart function remains stable and is doing very well developmentally. He never stops talking and singing. He is very observant; he doesn't miss a thing. We are very blessed to have Bly in our lives. We are also very fortunate to have wonderful family and friends to help and support us."*

*~ Tracy Brody*



# Barth Syndrome Trust - UK & Europe

By Michaela Damin, Chair

## The London Marathon – Sunday 23rd April 2006

This year we were fortunate to have two people running this gruelling marathon to raise funds for the Barth Syndrome Trust.

The first is Dave Allen, a friend of the Coleman family, who was so inspired by William, 16, who has Barth syndrome (see page 22), that he decided to give it his best shot. He made it in 4 hours 13 minutes 52 seconds and raised £4000 with a little help from the Coleman family. The money will be divided between BST and Great Ormond Street Hospital Children's Charity. Will's family hosted a dinner party with a successful auction at their local pub. Dave and I both needed an extra swig of wine before our speeches but we had a very generous audience with not one heckler! Thanks to the publicans and everyone who helped make this event such a huge success.

Dr. Honor Powrie is an Engineer at Smiths Aerospace and a colleague of Marco Damin, Nick's father. Nick is almost 8 years old and has Barth syndrome. Seeing how involved Marco and I are in the Barth Syndrome Trust, Honor decided that she too would seek sponsorship for the London Marathon. Honor completed the 26 miles in 4 hours 37 minutes and 28 seconds, and raised £1500. Thank you Honor!

## BST - Family gathering in The Netherlands, Europe on 22 April 2006

Four Dutch speaking families from Belgium and The Netherlands (5 Barth boys/young men, ages 18, 16, 13, 6 and 4) gathered at the Support-Expo in Utrecht in The Netherlands on Saturday 22 April 2006.



*Dutch speaking families renew friendships at Expo!*

The families met at a terrace in the centre of an exhibition about living with a chronic illness or disability. Many products and tools intended to make life easier, were shown at this exhibition. Many organisations offering services to people with a disability gave presentations.



*Chair-lift passes test.*

But the most important aspect for our Barth syndrome families during this day was just to meet each other in person, to put faces to names and to chat with each other about daily life with Barth syndrome. The boys had fun together. The parents shared their experiences. The meeting ended with a meal in a restaurant near the expo.

## Getting the word out across the UK - BST at Roadshows with the Children's Heart Federation (CHF)

The roadshows will feature an exhibition of services provided by CHF and its member groups. During the morning session the families will have the opportunity to meet and gather information about available services. The afternoon session will be a Professional Study Day for medical and other professionals.

The first roadshow is at Alder Hey Hospital, Liverpool on 15th June. Other venues for 2006 include Bristol, Birmingham and Northern Ireland. See [www.barthsyndrome.org.uk](http://www.barthsyndrome.org.uk).

## Creating new channels of communication – BST's Funding Programme encourages doctors from UK and Europe to attend 2006 BSF International Scientific, Medical and Family Conference.

Anyone who has been to a BSF conference can vouch for the fact that this is a unique event which gathers all the best minds, dedicated professionals and family members. It is an opportunity for doctors to share knowledge and meet the largest cohort of affected individuals ever gathered under one roof. In order to serve the needs of diagnosed (and as yet undiagnosed) cases here, we like to work with interested local doctors and scientists and encourage them to attend.

If you are a doctor or scientist working in the UK who has an interest in Barth syndrome, please contact the Barth Syndrome Trust. We will be happy to consider funding your travel and lodging expenses to our next conference, July 3-8th in Orlando, Florida. We would love to see you there.

*(Cont'd on page 21)*

(Cont'd from page 20)

### Calendar Girls Bare (Nearly) All for Barth

The Group Image Department at HSA in Andover, Hampshire, after reading about BST in their local press, created their own Calendar Girls type calendar. Thank you to Rebecca Curtis and colleagues for raising £550 and a laugh or two.



Your deal!

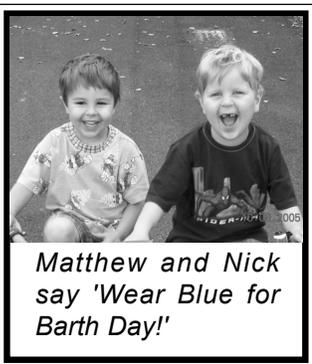
### Anne Ward's Bridge Party – 24th April – Rural Hampshire

Forty very keen bridge players arrived at the home of Maggie Barnard to be greeted with wine and elderflower cordial. A splendid lunch of salmon, new potatoes and salads followed by delicious puddings prepared by Mary Williams, fortified the players for an afternoon of Rubber Bridge.

There were many good parting comments. Most of our guests hadn't heard of Barth syndrome before but thanks to the information leaflets another forty people are aware of the syndrome, including doctors, doctors' wives and doctors' mothers. The event raised £600.

### Wear Blue for Barth Day – summer 2006

Nick, aged 7½ was recently chosen by his classmates to be their Charity Representative. The class representatives choose the charities the school will support for the year.



Matthew and Nick say 'Wear Blue for Barth Day!'

In true "Nick" fashion, he made a successful sales pitch for the Barth Syndrome Trust. The children of Halterworth Primary will have a "Wear Blue for Barth Day". They will all make a donation and dress up for the day. We will be distributing badges, lapel pins and balloons and we will be holding a cake sale in the playground. We'll keep you posted....

### Other News

Our Board of Trustees recently welcomed Sarah Bull as a trustee. Sarah has been involved in the Trust since two of her sons were diagnosed with Barth syndrome. We also welcome her husband, Dave who has agreed to join Family Services. You probably know Dave and Sarah from their fund raising events. Together they have raised over £8000 for the Trust.

<b>Barth Syndrome Trust</b>		
<b>Statement of Financial Position</b>		
<b>as of December 2005</b>		
<b>Assets</b>	<b>2005</b>	<b>2004</b>
Donations & Fundraising Activities	£22 905.00	£16 956.00
Expenses	(£5114.00)	(£3551.00)
<b>Net Assets</b>	<b>£17 791.00</b>	<b>£13 405.00</b>

We would also like to thank Nori Damin who has taken on the unenviable task of keeping our books. This leaves our Treasurer, Jerome Doherty-Bigara, with more time for his awareness efforts.

And lastly, we held our Volunteer Workshop on 18 March 2006. We spent a very productive day reviewing the efforts of the last 6 months and mapping our route for the year ahead. These workshops are always exhausting yet energising. It is wonderful to meet face to face and to see how each person's efforts make a real difference to the advancement of the Barth Syndrome Trust. Our next workshop is set for Saturday, 4th November 2006 in Hampshire. Please come and join us.



Barth Syndrome Trust also has representatives in the following regions:
<b>Northern Europe:</b> Mrs. Jo van Loo (Languages supported: Dutch, English, German) Pr. Brummelkampstraat 49 8191 XC Wapenveld THE NETHERLANDS jovanloo@barthsyndrome.org
<b>Southern Europe:</b> Mrs. Eva Antomarchi (Languages supported: French, Italian, English) Les Acacias Bâtiment B Quartier La Sèbe 18 chemin Sainte Thérèse 04 000 Digne – les - Bains FRANCE eantomarchi@barthsyndrome.org

# What Our Volunteers Are Saying...

By Terri Allison, BST Volunteer

I first became involved in helping with BST when a lady I met at a church service asked me if I would



help at a jumble sale. The lady was Annick Manton, mother of BST Chair Michaela Damin and grandmother of Nicholas (who has Barth syndrome) and Matthew.

As the jumble sale drew to a close, this bubbly chatty little boy blew into the hall and captivated us all.. Nicholas, 'my little charmer' had me hooked. Since that day in early 2004, I have been 'spreading the word' about Barth syndrome while trying to make as much money for the Trust as I can.

I grew up in Overton in Hampshire, where Annick now lives. At 19, I left home to work as a children's nanny in London. Although I loved looking after the children, it was very lonely and I moved on to work as a civilian in New Scotland Yard, (the Metropolitan Police Headquarters). Having married in 1985, I returned to live in Oakley (4 miles from Overton), in 1987. Two years later we were blessed with a son, Mark. I now work as a Learning Support Assistant at the junior school in Oakley.

The thought of the total isolation and despair that Michaela, Annick and family must have felt when Nick was diagnosed had a great impact on me. I doubt very much that I would have had the strength, determination, ability and sheer guts to start a Trust and challenge the world to

take notice of what 'my boy' is going through.

In the last eighteen months, with the help of the school, villagers and friends, the BST has become well known in Oakley and Overton, through numerous fundraising events. We have many more ideas and plans for the future.

My son, now 16, and his friend, Josh, have helped raise funds. They have designed tickets and posters, manned doors at discos, collected money at Christmas wearing silly antlers, and have generally been very supportive.

All children are precious and I'm proud to be part of a team that is actively pursuing every child's right to a happy and healthy life.



## Will Inspires Dave All the Way to the London Marathon

By William Coleman, England

My name is William Coleman and I'm 16 years old. I have had many problems in the



Catch of the day!

past with my health and am finally back at school full time. I am currently studying

hard for my GCSE's (examinations) which start in May.

During the Easter holidays I went up to London with my Mum, Helen, my Dad, Mike and my sister, Eleanor to

support my boss, Dave, who manages the shop where I work at weekends. He ran in the London Marathon to raise money for the Great Ormond Street Hospital Children's Charity and the Barth Syndrome Trust. (See page 20)

Dave has always wanted to run in the London Marathon and as soon as the chance came, he jumped at it. When he first applied to run just for the Barth Syndrome Foundation he could not get a place because there was a waiting list, so he applied to run for Great Ormond Street because they reserve 150

places for people to run for them. (I have been helped by Great Ormond Street many times over the years.)

'I like playing the guitar and also I have to admit I am a big fan of the Playstation. I am also a huge music fan and like bands like Pink Floyd, Dire Straights and the Red Hot Chilli Peppers - basically anything with a decent guitar part.'

Helen writes, "In July of last year Will was very surprised and delighted to be awarded the Diana, Princess of Wales Award for having succeeded against the odds. We are all very proud of him."



# Barth Syndrome Foundation of Canada 2005 ~ A Very Successful Year

By Lynn Elwood, President



*BSF of Canada Executive:  
(L-R) Carol Wilks, Cathy Ritter,  
Karen Gordon, Chris Hope, Lynn  
Elwood, and Lois Galbraith (not  
shown is Susan Hone).*

Each time we sit down to write a note about the events of BSF of Canada during the period since the last newsletter, we are surprised to see how much has happened in the last few months.

2005 was very successful. Our year ended with significantly more funds than we budgeted for. There were several contributing factors to this in the fall timeframe: successful fundraisers in the Golf Tournament and the Poinsettia sales, and some significant donations. We are very excited about the funding success in 2005 because it is allowing us to plan for expanded programs in 2006. We are now in a position to allocate funding for Scientific and Medical grants and are hopeful of finding a suitable applicant in the next year or two. We are able to help in funding some portions of the upcoming conference and we are able to attend some Awareness events. All of this is possible because of the contributions in both time and funds from many people.

During the fall we attended two Awareness conferences with the Barth Syndrome Foundation. We expect to continue these efforts in 2006 and are excited that Catharine

Ritter has received an invitation to speak at the fall Child Neurology Conference. This is a major accomplishment and will allow BSF to further awareness in the nursing community.

Some of us are involved in helping to organize portions of the 2006 International conference in Orlando. We are also working to convince Canadian families and physicians to attend this important event.

In April we held our second Annual General Meeting, in conjunction with our second Canadian Outreach meeting, in the Toronto area. It was nice to see many of the families at this event. Presented were our financials and information on the last and coming years. Thank you to Cathy Ritter and Karen Gordon, who have agreed to serve another term on our Board of Directors.

"Birth of Barth Month" in May promises to be a busy month for fundraising activity. Audrey Hintze and Tony Murphy are hosting their second musical variety show and fundraiser on May 7 in Markham, Ontario. We are also doing a mail campaign and collecting change for Barth again. Havelock IGA donated containers which will be given out for collecting change and we're looking forward to the rolling of coins at the end of the month. This is a fun event for all ages to participate. The next big fundraiser after May is the Second Annual Golf Tournament on September 11. Mark your calendars and plan to join us for this day of fun.

We have been very lucky to have received help in many forms from literally hundreds of people. Whenever we put together the Friends of Barth list we are impressed by the numbers of people and organizations that have provided time, advice, products, funds and support for our organization. Two of these people have agreed to take on formal roles in our Executive. Carol Wilks has joined us as our Accountant, and Susan Hone has joined us officially on the Executive. We are looking forward to working closely with both Carol and Susan and know the organization will benefit from their experience, wisdom and enthusiasm. Welcome to the team!

As you can probably tell from our updates, the Barth Syndrome Foundation of Canada is growing and changing each year. If you or anyone you know would like to be part of the team or have talent or knowledge you would like to share with us, please get in touch with us. The help is always welcome.



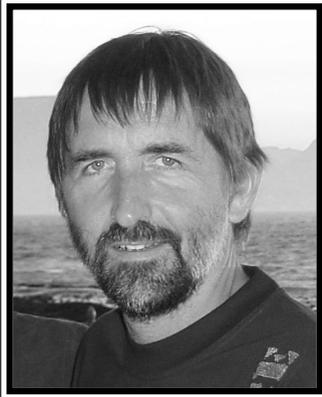
## The Barth Syndrome Foundation, Inc. Statement of Financial Position At December 31, 2005

	2005	2004
<b>Assets</b>		
Current		
Cash	\$ 38,010	\$ 7,675
Accounts receivable	\$ -	\$ 734
Prepaid expenses	\$ 965	\$ -
<b>Total Assets</b>	<b>\$ 38,975</b>	<b>\$ 8,409</b>
<b>Liabilities</b>		
Current		
Accounts payable & Accrued charges	\$ 1,103	\$ 600
<b>Fund Balance</b>		
Unrestricted	\$ 37,872	\$ 7,809
<b>Total Net Assets</b>	<b>\$ 38,975</b>	<b>\$ 8,409</b>

# First South African Physicians to Attend Conference Thanks to Barth Trust of South

By Jeannette Thorpe, Chair

I would like to take this opportunity to introduce two South African doctors who will be attending BSF's 2006 Scientific/Medical & Family Conference, and who are two of the representative doctors of Barth syndrome in South Africa.



**DR. RIK DE DECKER**

(MSc (cum laude); MBChB (Cape Town); DCH (RCP, London) : Diploma in Child Health; FCPaed (South Africa); Specialist qualification in Pediatrics; Subspecialist in Pediatric Cardiology; Cert

Med Genet (SA): subspecialist qualification in Medical Genetics).

Dr. De Decker is the senior specialist medical geneticist and a part-time pediatric cardiologist at the Red Cross Children's Hospital, Cape Town, South Africa.

His main interest is in cardiogenetics (the interface between genetics and congenital heart disease). Dr. De Decker also has an interest in medical teaching and computer resources for medical education and, as such, is the Chairman of the Red Cross Hospital Learning Committee.

When he has time, he pursues his love of mountaineering and is the co-convenor of the Cape Town section of the Mountain Club of South Africa Mountain Rescue Team, as well as the Wilderness Search and Rescue medical workgroup. In fact, after the medical conference in the USA, he and his family plan to climb in Lake Tahoe and Tuolomne Meadows in the Rockies (if anyone cares to join them!!).



**DR. JEFFREY HARRISBERG**

(M.B.B.Ch (Wits), FCP (SA), DCH, DTM&H. Specialist qualification in Pediatrics and subspecialist in Pediatric Cardiology).

Dr. Harrisberg was a full-time consultant at the Johannesburg Hospital for three years (and then part-time for five years). He is now involved, on a full-time basis, in a group private practice at Sunninghill Clinic, Gauteng. This practice is a referral center for many countries north of our borders (Botswana, Angola, Malawi and Zambia).

Between 2001 and 2003, Dr. Harrisberg was the President of the Pediatric Cardiology Society of South Africa. He has a particular interest in interventional cardiac catheterizations (e.g., valvuloplasties, ASD, PDA, VSD occlusions, intracardiac stents). He also has an interest in fetal cardiac ultrasound (diagnosing and managing congenital cardiac lesions antenatally).

Dr. Harrisberg is one of the doctors that has been looking after the only two known cases of Barth syndrome in South Africa for the past 10 years, and has observed the many ups and downs that this disorder presents.

When Dr. Harrisberg has time off, he enjoys spending time in the tranquil African bush and is a keen bird watcher.

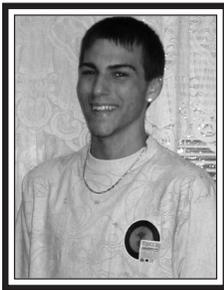
I would like to thank Drs. De Decker and Harrisberg for their support and interest in Barth syndrome and for taking time out of their busy schedules to attend our Medical Conference - including the marathon trip from South Africa to the USA! I look forward to working with them on educating people in South Africa about Barth syndrome.



# Barth Syndrome Sibling Spotlight!

By Jess Wiederspan, Nebraska

Below are the profiles of four of our fantastic BSF siblings. Please e-mail Jess Wiederspan at [onionhater1979@yahoo.com](mailto:onionhater1979@yahoo.com) with questions or comments.



Justin

**Age:** 16  
**Name and age of Barth Sibling:** Derek, 12  
**Where do you currently live?** Altamont, New York  
**What grade are you in?** 10th  
**Career goals:** I want to be a professional soccer player  
**Favorite Movie:** Waiting  
**Favorite TV Show:** Lost  
**Favorite Book:** Stormbreaker  
**Favorite Food:** Italian & Seafood  
**Hobbies:** I enjoy soccer, snowboarding, rollerblading & working  
**Famous person I'd most like to have dinner with:** David Beckham  
**Something unique about me is:** I enjoy mechanical drawing, am very good at math, and enjoy helping my friends with their math studying and homework.  
**As a Barth sibling, I think the best thing about the Barth Syndrome Foundation is:** It is helping to find a cure!



English

**Full Name:** Taylor English  
**Age:** 13  
**Name and age of Barth Sibling:** Benjamin, 9  
**Where do you currently live?** Knoxville, Tennessee  
**What grade are you in?** 7th  
**Career goals:** Going to college to study acting or singing  
**Favorite Movie:** She's the Man  
**Favorite TV Show:** Drake and Josh, American Idol  
**Favorite Book:** Number the Stars  
**Favorite Food:** Chicken Alfredo  
**Hobbies:** playing softball, basketball, swimming, singing and acting!  
**Famous person I'd most like to have dinner with:** Kelly Clarkson  
**Something unique about me is:** My name!  
**As a Barth sibling, I think the best thing about the Barth Syndrome Foundation is:** We get to meet people from all around the world and make new friends. BSF also helps doctors keep my brother healthy.



Justin

**Age:** 9  
**Name and age of Barth Sibling:** Adam, 16  
**Where do you currently live?** Ajax, Ontario  
**What grade are you in?** 3rd  
**Career goals:** I would like to be in the army and protect my country.  
**Favorite Movie:** Pirates of the Caribbean  
**Favorite TV Show:** Inuasha (Japanese Anime)  
**Favorite Book:** Deltora  
**Favorite Food:** Spaghetti & Meatballs  
**Hobbies:** Hockey, Lacrosse, Swimming, Computer Games, Card Games, Hide and go Seek  
**Famous person I'd most like to have dinner with:** Johnny Depp  
**Something unique about me is:** I like to spike my hair and I have the neatest bedroom in the house.  
**As a Barth sibling, I think the best thing about the Barth Syndrome Foundation is:** They are so willing to help the people who need help and that is so kind.



Jessica

**Age:** 26  
**Name and age of Barth Sibling:** John, 24  
**Where do you currently live?** Lincoln, Nebraska  
**What grade are you in?** I currently work as a grant writer for a non-profit agency and will begin working on my joint Ph.D. in Social Work and Sociology at the University of Michigan in the fall.  
**Career goals:** I plan to teach at a college or university & research issues related to poverty.  
**Favorite Movie:** The Sound of Music  
**Favorite TV Show:** The Daily Show, American Idol  
**Favorite Book:** Savage Inequalities by Jonathan Kozol  
**Favorite Food:** Indian food (saag paneer)  
**Hobbies:** Reading, writing, painting, running.  
**Famous person I'd most like to have dinner with:** Jon Stewart and/or Jonathan Kozol  
**Something unique about me is:** I have been a vegetarian for the past 10 years but I hate lettuce.

# Power of Kindness ... Contributions received January 2005 to present

## Science/Medicine Fund

**Angels**  
 Anonymous  
 Bailey, Clarke/Trish  
 Barad, Seth/Amy  
 Blumenthal, Richard/Cynthia  
 Bogert, Carroll  
 Buly, Dr. Robert/Lynne  
 Campbell, Tony/Debbie  
 Cusack, Tom/Carrie  
 Geary, Ann  
 Hart, Dana  
 Lummis, Dr. Fred/Marliyn  
 Malkin, Mr./Mrs. Peter (The Malkin Fund)  
 Malkin, Scott/Laura  
 McCurdy, Steve/Kate  
 Rodbell, Arthur/Rhonda  
 Russell, Sara/Paul  
 Russell, Dr. Paul/Alene  
 Shapiro, Earl/Brenda

## Contributions of \$50 and Above

Brown, M. Vanburen  
 Dispatch Transportation Corp.  
 Garrity, Jonathan /Caroline  
 Hazen, Edward/Elizabeth  
 Holly, Peggy M.  
 Leon, Susan (The Leon/Fredman Family)  
 Lu, Vincent/Kristen  
 McFee, Wendy  
 Randolph, Dr. Peter/Helen  
 Rubin, Dr. Nina/Thomas  
 Schlosser, Alfred/Jacqueline  
 Sherer, Peter/Marilu  
 Taussig, Tim/Nancy  
 Watt, Richard/Gillian

## Woody Varner Science/Medicine Fund

**Angels**  
 Abel Foundation  
 Brehm, Russell/Louise  
 Budig, Gene/Gretchen  
 Cheatham, Dr. John/Mrs.  
 Dillon Foundation  
 Koeftot, Dr. Bob/Mrs.  
 Richard & Virginia Voell Family Fund  
 Schorr, Paul/June  
 Swanson Biggs Adams Family  
 Foundation)  
 The Pegler Family Foundation  
 Tinstman, Dale/Jean  
 Varner, Paula  
 Wilkins, Dr. Mike/Sue  
 Yanney, Mike/Gail

## Contributions of \$50 and Above

Allman, Peter/Maureen  
 Allman, Thomas E.  
 Basler, Dr. Rod/Debbie  
 Buckley, Les/Nancy  
 Beynon, Dave/Liz  
 Bingham, Dr. Dave/Kathy  
 Burmeister, Charles/Marita  
 Campbell, Bob/Candy  
 Dimon, Gerry/Kit  
 Dittman, Alice  
 Fairfield, Terry  
 Farrar, Doug/Shawn  
 Faulkner, Mrs. Clarke  
 Firestone, Dave/Jane  
 Gelber, Dr. Ben/Elaine  
 Hedgecock, Norm/Debbie  
 Henricks, Dr. Bruce/Peggy  
 Hurlbut, Dr. Greg/Sheri  
 Kenyon, Barbara/Allan  
 Kiechel, Dr. Fred/Vivian  
 Kingery, Orville/Dorothy  
 Lacy, Dr. Sushil/Jane  
 Lauerman, Jim/Meg  
 Lehr Family Trust  
 Linder, Dr. Max/Pat  
 Massengale, Dr. Martin/Ruth  
 McClendon, Joe/Lynn  
 Minnick, Gates/Daisy  
 Nelson, Scott/Teri  
 Norris, Dr. Mike/Mary  
 Olson, Loy/Julie  
 Osborne, Rep. Tom/Nancy  
 Otte, Rob/Carolyn  
 Perlman, Chancellor Harvey/Susan  
 Pederson, Steven/Tami  
 Pittenger, Jim/Julie  
 Rawley, Ann  
 Roskens, Dr. Ron/Lois  
 Roth, Dr. Bob/Marcia  
 Sample, Dr. Steve/Catherine  
 Seacrest, Gary/Sue  
 Seacrest, Jim/Ronda  
 Seaman, Andrew/Steely, Robyn  
 Shamblin, Dave/Sue  
 Sonderegger, Ted/Mary Ann  
 Spurrier, Hal/Nancy  
 Steely, Robyn  
 Stohs, Dr. Gene/Kristen  
 Stuart Foundation

Stuart, Jr., James  
 Stuckey, Dennis/Nancy  
 Tegt, Dr. Tom/Barb  
 Varner, Judy  
 Varner, Tom/Beth  
 Waring, Dr. Tom/Mrs.  
 Weesner, Dr. James  
 Whitehead, Bus/Jeanne  
 Wilkins, Dr. Lee/Kristi  
 Wilkins, E. Joanne

## General Fund

**Angels**  
 Bauer Family Foundation  
 Bullock, Betty  
 Chenault, Kenneth (The Ayco Charitable  
 Foundation)  
 Crowley, Peter/Gretchen  
 Davis, Martha  
 Fowler, David/Susan  
 Johnson, Deloris/Malcolm  
 Kavetas, Mrs. Harry (The Harry L.  
 Kavetas Family Fund)  
 Kelly, Al/Peggy  
 Lake City International Trucks, Inc.  
 Lindsey, Mr./Mrs. John (Sara and John  
 Lindsey Foundation)  
 Mahoney, David/Winn  
 McCurdy, Steve/Kate  
 Monetti, Dorothy/Robert  
 Osnos, Susan/Peter  
 Pagano, James (Fully Involved Lawn  
 Service)  
 Papone, Aldo/Sandra  
 The Frill Foundation  
 The Lebensfeld Foundation  
 United Space Alliance  
 Vaisman, Beth/Natan  
 Wal-Mart Foundation  
 Winston, Laura

## Contributions of \$50 and Above

Anonymous  
 1507386 Ontario Ltd.  
 AAMCO Transmission of Merritt Island  
 Abbonizio, Leita  
 Acklie Charitable Foundation  
 Adams, Lynda/Phillip  
 Adams, Theodore/Alma Mills  
 Adams, Robert/Catherine  
 Adams, Sandra  
 Adapt Aviation Inc.  
 Aim Trimark  
 Aircraft Maintenance Engineers Assoc.  
 American Express Foundation  
 American Express Gift Matching  
 Anchor Road Stores  
 Anderson, Debra  
 Anderson, Frederick/Ruth  
 John Anderson and all at British  
 Aerospace, Prestwick  
 Andrews, David/Ellen  
 Antomarchi, Eva/Nicolas  
 Archer's Eyewear  
 Architects in Association  
 Arth, Lawrence  
 Asaf, Ernest/Colette  
 Aviall Canada Ltd.  
 BMW Canada Inc.  
 Baffa, Ted/Rosemary  
 Barbier, Etienne  
 Barrie Honda  
 Barrs Court School Fund  
 Bater, Jennifer  
 Beyel Bros. Crane  
 BD Corporation  
 Beefy Gang LLC  
 Benevolent and Protective Order of Elks  
 Berens, Wayne/Harriet  
 Bergquist, Clay  
 Berman, Carol/Walter  
 Berry, Rick/Carla  
 Beyel, Danny  
 Bez, Ann  
 Bialo, Katherine/Kenneth  
 Bishop Shanahan High School  
 Black's Spray Service, Inc.  
 Blaise Automobile Detail, Inc.  
 Block, David/Sharon  
 Blumenthal, Martin/Sally  
 Bob Steele Chevrolet, Inc.  
 Bock, David/Amy  
 Bogert, Nicholas/Sally  
 Boris, Marc  
 Bowen, Shelley/Michael  
 Boyere, Anne  
 Brabbs, Kelly/Madelyn  
 Bradley, Robert/Margaret  
 Bradley, Robert/Nancy  
 Bradt, Meg  
 Brenner, Andrew/Kathleen  
 Brevard County Pro. Firefighters  
 Brewer, Gerilyn  
 Bridger, Dianne/Wayne  
 Brophy, Thomas

Broughan, Malcolm  
 Brown & Brown, Inc.  
 Buckeye Community Federal Credit Union  
 Buckeye Florida, L.P.  
 Buddemeyer, Donna  
 Burdett, Alfred/Norma  
 Bull, Corey/Kai/ Ashley  
 Bull, Lyn  
 Bull, Sarah/Dave  
 Burns, Geraldine  
 Burns, Edward  
 Burtis, Cleo  
 Bussen-Mayer Engineering  
 Byrd, Todd/Michelle  
 Carroll, Ann/Robert  
 Case, Jr., Charles  
 Carveth, Dr. Steve/Beth  
 Cavanaugh, Matthew/Alice  
 CB Richard Ellis  
 Chandler, Cameron  
 Chapin, Lee/Barb  
 Chodock, Robert  
 Chuck's Lawn Service & Landscaping Inc.  
 Cingular - Matching Gift Center  
 Coleman, Helen/Michael  
 Collazzi, J.  
 Community Health Charities of California  
 Concannon, Maura/Richard  
 Condliffe, David/Cynthia  
 Condliffe, James  
 Coupland Insulation  
 Coutinho, Alexandra  
 Country Meat Cuts  
 Cox, Patricia/Gerald  
 Cruise Holidays of Barrie  
 Considine, Kate  
 Cook, Christopher  
 Corrow, Julie  
 Cracchiolo, James  
 Crowley, Charles/Diane  
 Culton, John/Patricia  
 Cunniffe, Francis/Helen  
 Curry, Jones T.  
 Damin, Mandy/Claudio  
 Dannels, Richard/Theresa  
 Davies, Stephen  
 Day, Margaret  
 Deforest, Ina/ Wilmer  
 Deforest, Lorraine/Dupont, Kevin  
 Deeter, Doug/Leslie  
 Dickie, Paul  
 Direct Line Sales  
 Dittman, Alice  
 Dix, Glenda/Lee  
 Dobrosky, Thomas/Paula  
 Dombesk, Mary Ann  
 Dowden, Carroll  
 Dowden Health Media  
 Doyle, Patrick  
 Drinkwater, Paul  
 Dungan, Robert  
 E J Berry  
 Eda, Nancy  
 Edwards, Bobby  
 Elmer, Keith (Youngsters Ltd.)  
 Elwood, Lynn/Rick  
 Elwood, Susan/Brian  
 Emerick, Douglas (Doug Emerick's Lawn  
 Svc.)  
 Emerson, Saffrone  
 Emery, Jonathan/Suzanne  
 Enos, James/Cheryl  
 Esther Gemell and all at the Coach and  
 Horses Pub  
 Evans, Robert  
 Fairfield, Terry  
 Falcon's Roost, Inc.  
 Farrell, Susan/Tim  
 Fenn, Greg  
 Fields, Wendell  
 Fink, Penny  
 Firestone, Dave/Jane  
 Firestone, James/Ann  
 Fleet Matching Gifts Program  
 Fletcher, James/Darwyn  
 Florence, David/Elizabeth  
 Flynn, Clare  
 Foster, Frank/Kathleen  
 Fowler, R. Dean  
 Franklin, Peter/Dorothy  
 Fraternal Order of Eagles #4257  
 Frost, Gemma  
 Frost, Michael/Angie  
 Frost, Tracy  
 Fuller, Russell/Judith  
 Gagnon, Lou Ann  
 Galbraith, Lois/Morris, Les  
 Gardiner, Timothy/Nancy  
 Garrison, Howard/Michele  
 Geeser, Gerald  
 Gemell, David and all at Ayr Hospital  
 Genetic Interest Group  
 Genthner, Harold  
 Geroux, Roxanne  
 Gilmour, Lisa/Mark

Glenn, Bill  
 Goodwin, William/Virginia  
 Grabarz, Jillian  
 Grant, Alec/Catherine  
 Graper, Teddy/Maureen Bird-Graper  
 Greene, Roy  
 Gregory, Karen  
 Gress, Emil/Natalie  
 Gunderson, Kent/Barbara  
 Gurr, Clifton/Sheila  
 Guthorn, Robert  
 HAS/Group Image Dept  
 Haas, Rod/Wendy  
 Hackett, Muff/Doug  
 Haessler, John/Nancy  
 Hall, Gregory/Candace  
 Hall's Mark of Brevard, Inc.  
 Hammett, Richard  
 Hampton, Mary  
 Hardison, Charles/Jodi  
 Harris, Jesse  
 Harris, John/Syble  
 Hartnett Properties  
 Hazen, Edward/Elizabeth  
 Health First  
 Heinley, Kyle/Ruthanne  
 Helen Keller Elementary School  
 Higgins, Liz/John  
 Hill, Mark/Sally  
 Hille, Jim/Tina  
 Hintze, Audrey  
 Hirschfeld, Paul  
 Hobbs Pharmacy United, Inc.  
 Hock Your Rocks  
 Holland, Mark/Diane  
 Hollowell, Steven/Robbin  
 Hone, Chris/Susan  
 Hope Aero Propeller & Components Inc.  
 Hope, Christiane/Michael  
 Hope, Harry/Helen  
 Howell, David/Patrica  
 Hudson, Thomas/Nancy  
 Hughes, Ronn/Cathy  
 If It's Water, Inc.  
 Irene Development Inc.  
 Island Sports Shack  
 Island Surf and Skate LLC  
 James C. & Rhonda S. Seacrest  
 Foundation  
 Jamson Visions, LLC  
 Jaspers, Valerie  
 Johnson, Linda  
 Jones, Edmunds & Associates  
 Jones, SE/Customs & Excise Wales  
 Joyce, David/Jane  
 Kalange, John/Teresa  
 Karp, Wendy/Matthew  
 Kearns, Richard  
 Keating, Barbara/Con  
 Kenney, Linda  
 Kew Beach Couples Club  
 Kenney, David (Digitas)  
 Kingery, Dorothy  
 Kinsella, Robert  
 Kirk Palmer & Associates, Inc.  
 Kirkham, D. Collier/Ann  
 Kittleson, Clay/Holly  
 Kiwanis Club of Perry, Inc.  
 Knowlson, Mr. E./Mrs. V.  
 Knudsen, Richard/Sally  
 Kordel, Chester/Rita  
 Kratchman, Joan/Robert  
 King, Dr. H. Roderic  
 Kugelmann, Steve/Jan  
 Kugelmann, Mike/Catherine  
 Kugelmann, Brian/Julie  
 Kugelmann, Mary Irene  
 Kugelmann, Tom/Hanh  
 Kugelmann, Warren/Maria  
 Kugelmann, Peter/Karen  
 Kugelmann Land Surveying, Inc.  
 Kunkle, Jr., John N.  
 L'aiken - A South Carolina Partnership  
 Labelcraft Products Ltd.  
 Lau, Tony  
 Lawson, D.C. Richard  
 Lawson, Doreen  
 Lee, John/Marguerite  
 Lever, Charles/Beverly  
 Lichte, Eric/Marie  
 Lincoln Community Foundation  
 Linen, Jonathan  
 Lind, Michael  
 Lochner, Suzanne  
 Lockhart, Paula  
 Lockhead Martin  
 Loggins Realty, Inc.  
 London Law Trust  
 Lorenc, Diane  
 Louis Rothschild Insurance Agency  
 Lyon, Robert  
 Lyon, Jeffrey  
 M2 Constructors, Inc.  
 Mack, Jonathan  
 Mancino, Rosemary/Angelo

Marks, Henry/Alexandra  
 Mann, David/Sheila  
 Marsh, Bette  
 Martinez, Dr. Jose/Deborah  
 Masterson, Moira  
 Matthias, Michael/Linda  
 McAleer, Rose Mary  
 McConaughy, James/Beverly  
 McCurdy, Chris/Kris  
 McCurdy, William/Virginia  
 McCurdy, John/Sharon  
 McCurdy, Steve  
 McDonnell, Diane  
 McElroy, John/Susan  
 McGill, Steve/Nancy  
 McGuinness, Christine  
 McJannett, Susan/Bob  
 McQuaig, Michael/Brenda  
 Medici, Jim/Christine  
 Meighan, Paula  
 Merchant's Financial Guardian Inc.  
 Merritt Island Lodge #2073  
 Met-Con, Inc.  
 Michaud, Steven/Yoko  
 Mike Erdman Motors, Inc.  
 Miller, Gary/Cornelia  
 Miller, Michael/Romaine  
 Miranda, Jr., John  
 Mitchell, Robin  
 Mixer, Stephen/Elizabeth  
 Monahan, Tom/Laurie  
 Moncure, Suzanne  
 Moncure, John  
 Montanaro, Louis/Theresa  
 Morehouse, L. Clark/Susan  
 Morganstern, Marc/Louise  
 Morris, Ian  
 Motzkin, Robert  
 Mueller, Henry  
 Murphy, Donald/Jane  
 Murphy, Tony  
 Myers, Judith  
 Naraine, Christopher  
 Nature Coast Inc.  
 Newman, Randy  
 Nicoll, Thomas/Mary  
 Nissen, James/Ginger  
 Noddle, Norman/Bev McKee  
 Northrup, Bruce/Kathryn  
 Novopharm  
 Oldewage, Scott/Casie  
 Olson, Richard/Sharon  
 Olson, Ken/Tina  
 Olson, Tom/Maria  
 Optimist Club of Perry Florida  
 Ordonez, Albert/Kimberly  
 Osterhout, Alfred/Melony  
 Owens, Pete  
 PQ Corporation  
 Paczkowski, Taisia  
 Paolizzi, Vincent/Celeste  
 Paradise Ford  
 Parker, Greg  
 Patroske, Pat (Ford Motor Co.)  
 Patrick, Barbara  
 Perkins, Phyllis  
 Peterson, George/Harriet  
 Pepsi Cola Bottling Group  
 Perini Building Company, Inc.  
 Perlman, Harvey/Susan  
 Perry Woman's Club, Inc.  
 Perry-Taylor County Chamber of  
 Commerce  
 Pfizer Foundation Matching Gifts Program  
 Philip, Katharine  
 Piranha Pools, Inc.  
 Pitkethly, Maureen  
 Poitras, Sandra/Michael  
 Pollack, Stephen/Judann  
 Precision Aero  
 Pringle, Matthew/Julie  
 Purcell, Robert/Jacqueline  
 Quickfall, Adrian  
 Rader, Stephanie  
 Rader, Carol  
 Real Time Lead Generation  
 Reed, Kathryn  
 Reilly, Dennis/Deanna-lyn  
 Reilly's Screen Enclosures  
 Reynolds, Suzanne  
 Reynolds, Smith and Hills, Inc.  
 Rigney, John  
 Rios, Alexander  
 Ritter, Carole  
 Roberts, John/Elizabeth  
 Robertson, Harriet  
 Rodan, Amnon/Katie  
 Rogers, Jimmy/Susan  
 Rogers, Lawrence/Catherine  
 Rosiek, Paul/Cheryl  
 Roskens, Ronald/Lois  
 Rothschild, Stephen  
 Rothschild, Adam/Katherine  
 Runaway Investments, Inc.  
 Russell, Harold/Margo

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## Contributions of Time and Advice

Adelstein, Dr. Robert  
Allison, Mark  
Allison, Terri  
Anderson, Suzie  
Antomarchi, Nicolas/Eva  
Ashenfarb, David (Schall & Ashenfarb)  
Baffa, Mary  
Baffa, Rosemary  
Bellamy, Amanda  
Barth, Dr. Peter  
Berry, Carla

# BSF Gets a New Website!

Lynn Elwood, Chair, BSF Technology Committee

As you know the Barth Syndrome Foundation website [www.barthsyndrome.org](http://www.barthsyndrome.org) has been a tremendous source of information of all types for all audiences for a number of years. There are hundreds of pages of information on the site and there are many visitors to it every month.



Over the last 6 months there has been a project underway to recreate the website with a new look, an organization more reflective of all our audiences, and greater ease of finding material. By the time this newsletter arrives, the new website should be launched and you should be seeing the new [www.barthsyndrome.org](http://www.barthsyndrome.org).

On the new site there are categories for each audience, easy paths to the information people are looking for and a comprehensive search to find information throughout the site. There is more information available in different languages, and links to sites of all the affiliate organizations.

One of the advantages of the new website is that a variety of people can provide updates so the content will be much easier to keep current. There was quite a team of people involved in putting all the content together and creating this new site. First there is Pixelera, the Ottawa-based company (recommended through the Genetic Alliance) who created the

artwork for the site, will host it and has guided us through every step of the project. There were also the program leaders who have provided information for sections of the site. Then there was the core team that put together most of the pages, the links, the email addresses and made the project come together. Thanks to everyone for all their hard work on this and special thanks to Lynda Sedefian, Michael Hope, Joke van Loo, Michaela Damin, and Shelley Bowen, who were the source of so much on the original website.

Please visit [www.barthsyndrome.org](http://www.barthsyndrome.org) often and let us know if you want to see something you don't find there. This is a dynamic site that will grow with time and your ideas.

*Note: BSF would like to publicly thank Lynn Elwood for her valued time and talents in spearheading the transformation of BSF's new website.*

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



Barth Syndrome  
Foundation

The Barth Syndrome  
Foundation, Inc.  
P.O. Box 974  
Perry, Florida 32348

Phone: (850) 223-1128  
Facsimile: (850) 223-3911  
E-mail: [info@barthsyndrome.org](mailto:info@barthsyndrome.org)