



Barth Syndrome  
Foundation

www.barthsyndrome.org

# NEWSLETTER

Volume 7, Issue 1

\$0.80 cents USA

## 2006: A year of great progress for BSF



*By Stephen McCurdy, Chairman and CFO and Shelley Bowen, President*

**Marking the previous year's progress in our Spring Newsletter has become a point of pride and this year is no exception.** We are pleased to report the most successful BSF Conference ever, the initiation of the long awaited Barth Syndrome Medical Database and BioRepository, an entirely re-designed website, more families enrolled, more research grants awarded, near record fundraising and the hiring of our Science Director – Matt Toth, PhD - virtually all accomplished by a dedicated group of volunteers. In the article that follows, you will get a taste of BSF, our programs, our volunteer leaders and our financial health. But our pride is always tempered by the knowledge that our goal is still very distant... and boys are turning into young men without a solution to Barth syndrome in hand. Thanks to each of you and your continued willingness to help, we remain optimistic. Our hope for the future rests with you.

### BSF PROGRAMS' PROGRESS

#### *FAMILY SERVICES:*

#### **SHELIA MANN AND CHRIS HOPE, CO-PROGRAM LEADS**

We continue to serve our community of families around the world through our internet listserv which allows every family (including the boys and young men themselves) to speak to each other regularly. Families can ask questions, share milestones and provide reassurance that we are not alone in this sometimes overwhelming and frightening journey. In addition to providing educational materials to the families, our Family Service team also provides peer-reviewed information about Barth syndrome, which families can share with their physicians.

*(Cont'd on page 3)*

## BSF is developing a global roadmap for Barth syndrome research

*By Matthew J. Toth, PhD, Science Director, BSF*

**Our 2006 Research Grant Program has just been completed and has awarded almost \$320,000 in funding to eight applicants, a record for a single grant cycle.** The projects funded range from basic research into the interactions of the tafazzin gene product (the gene mutated in Barth syndrome) with other genes/proteins to clinical projects that will measure biochemical levels, biophysical

properties, and mental acuties of boys with Barth syndrome. This grant cycle is particularly rich with projects that seek to build on the work previously funded by the BSF, which indicates that progress in Barth syndrome research is scientifically sustainable and rewarding. Brief descriptions of the funded projects are listed on page 11.

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Children's Hospital  
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Neurology  
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Molecular Diagnostics Lab  
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A. I. DuPont Hospital for Children  
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**Miriam L. Greenberg, PhD**

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Anesthesiology  
NYU School of Medicine  
New York, New York

**Colin G. Steward, FRCP,  
FRCPCH, PhD**

Pediatric Hematology  
Bristol Royal Hospital for Children  
Bristol, England

**Jeffrey A. Towbin, MD**

Pediatric Cardiology  
Texas Children's Hospital  
Baylor College of Medicine  
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**Ronald J. A. Wanders, PhD**

Genetic Metabolic Diseases  
Academic Medical Center  
Amsterdam, The Netherlands

**Katherine R. McCurdy**

*– ex officio*  
Science and Medicine  
Barth Syndrome Foundation, Inc.  
Larchmont, New York

**Matthew J. Toth, PhD**

*– ex officio*  
Science Director  
Barth Syndrome Foundation, Inc.  
Iselin, New Jersey

# Special gifts

## **By Valerie ("Shelley") Bowen, President**

In 2001, we celebrated the creation of the Barth Syndrome Foundation - the first charity to aid those who have Barth syndrome. Our mission was to serve a community with significant needs: Barth syndrome was claiming lives, constantly threatening the health of those with the disorder and an ever-present challenge to the families and physicians caring for them. On behalf of the community, we vowed to do everything in our power to change the status quo and bring hope to those affected by Barth syndrome, and we felt accountable to deliver on our promise.

This edition of our Newsletter provides an account of our efforts toward our vision ... ***"Saving lives through education, advances in treatment and pursuit of a cure for Barth syndrome."*** It reports on the effectiveness of our organization as we consider the milestones and accomplishments we have reached. Since our founding, BSF has surpassed many of our initial goals and is transforming the future of those who have Barth syndrome.

Over the years, appreciating the urgency of our mission, volunteers and donors have selflessly given their time, skills and funds to BSF. We have tried to be good stewards of the time, talent and treasure entrusted to us, investing wisely in our programs and planning for the future. But in the end, though we track our milestones, the real measure of our success is more human – our ability to improve the lives and futures of our boys, many of whom are now becoming young men.

It is impossible to measure the worth of a life that is lived and loved. It is equally impossible to measure the depth of our gratitude toward those who make a difference in our loved one's life. The tenderness of a touch, the sound of a child's voice, the smell of his hair and the caress of his fingers as we read bedtime stories are the real gifts you all give to us. These are ordinary happenings in most families. But for our families, they are gifts made sweeter by the threat of Barth syndrome. We are a unique community that Barth syndrome has forever defined and changed. We are an extended family separated by thousands of miles, who care deeply for one another and for each other's children. Strong friendships have formed, which are the core of our culture. These are the unexpected gifts on our journey.

My aspiration throughout childhood was to be a mom. Watching my mother raise my brother, an atypical child with special needs, she inspired me as a quiet model of strength, independent thought and determination. Little did I know that I would draw upon these unspoken lessons when I had children of my own.

From my mother I learned that special needs children are not broken or damaged. My brother's challenges would enrich, not defeat our family. To this day, his perspective brings joy to all who know him. I have often heard him refer to those undeserving of his kindness as his 'friends'. Through him I have learned the true meaning of compassion. When we deeply care for someone they are a part of us. I was reminded of this simple truth when a young boy in our group said, *"I am affected by Barth syndrome because my brother has it."*

There are no words to ease the unbearable pain and grief a parent experiences with the loss of a child. However, the amazing gifts that these children leave with us are imprinted on our souls. Their lives and our love for them will never be forgotten. I choose not to remember the loss of one of my sons, but to be inspired by the joy he brought into my life.

In 2006 our greatest victory of all was that not one family in our group experienced the loss of a child. It is difficult for me to convey the gratitude I have for the investments made in BSF by so many so that ... ***"no one will suffer or perish from Barth syndrome."*** There are no words to express the depth of gratitude for those who have elected to join us in our journey, and there are no measures for unconditional friendships.

None of us planned on being here and none of us has a map. We must create our own solutions. But as unexpected is our membership in this unique community, so too are the partners, like you, whom we find at our sides and with whom we will find the answers we need.

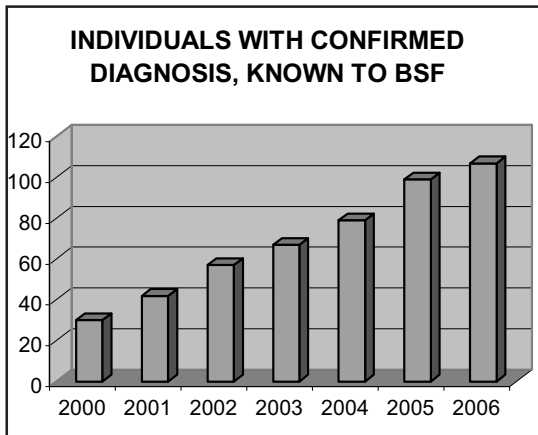


2006 ~

# A year of great progress for BSF

(Cont'd from Cover)

This is a credible and invaluable resource for these clinicians. Families (and physicians and scientists) can also join BSF at various regional outreach meetings held throughout the US, Canada, the UK, Europe and South Africa. BSF's next outreach meeting will be held in Brockton, MA on August 17-20, 2007. For more information, please visit our website at [www.barthsyndrome.org](http://www.barthsyndrome.org).



At the end of 2006, Family Services was working closely with the families of 107 individuals living with Barth syndrome and an additional 22 families awaiting diagnosis. Among those we now serve are 13 affected individuals over the age of 20, one of whom is 45 ... A remarkable demographic in comparison to the early data which described BTHS as a disorder fatal during infancy! Our support services are changing to accommodate the concerns of young men, some of whom have families of their own.

## **BSF CONFERENCE: JAN KUGELMANN, PROGRAM LEAD**

2006 was a Conference year for BSF (we hold international conferences with tracks for scientists, physicians and families every two years). Our last Newsletter detailed the great success of the Third International BSF Scientific, Medical and Family Conference held in July 2006. Our Conference is one of the centerpieces of BSF's programs, as it brings our many research grant recipients together with scientists from premier research institutions around the world. It joins the science of the researchers with the bedside experience of the physicians attending our boys to develop and document improved treatment protocols. BSF's Conference also brings the many affected families together to attend two days of medical clinics and discussions on the progress made by the scientists. This increased knowledge about the complex, multi-system disorder the families face daily is critical. There also is a chance to connect with other families – their fellow voyagers on a lifetime trip for which none had made a reservation. These connections are invaluable as they build trust, lend crucial emotional and practical support, cement what might otherwise be unlikely friendships, and re-charge our collective enthusiasm for the work ahead.

## **SCIENCE AND MEDICINE: MATTHEW J. TOH, PHD, BSF SCIENCE DIRECTOR**

### **Professional Science Director Hired**

As has already been reported in a previous newsletter, one of the great advances made by BSF in 2006 was the hiring of a professional Science Director to oversee and further stimulate all of our science and research programs. In an effort led by Kate McCurdy, Matthew J. Toth, PhD was recruited and began with BSF in July 2006. Matt has a PhD in microbiology from MIT and nearly 20 years of very relevant experience in the pharmaceutical industry. He has already proven just how beneficial it is to have someone with his background and perspective on our team full-time, and he also has shown repeatedly that his personal qualities are a perfect fit with BSF as well. We are truly delighted to have him with us, and we know we will continue to see the very positive results of his involvement.

(Cont'd on page 4)

For all correspondence, including information, please contact:

#### Board of Directors

Valerie (Shelley) Bowen, President  
sbowen@barthsyndrome.org

Michaela Damin, Board Member  
mdamin@barthsyndrome.org

Stephen Kugelmann, Awareness  
skugelmann@barthsyndrome.org

Katherine R. McCurdy, Science/Medicine  
kmccurdy@barthsyndrome.org

Stephen B. McCurdy, Chairman & CFO  
smccurdy@barthsyndrome.org

Susan S. Osnos, Board Member  
sosnos@barthsyndrome.org

Susan V. Wilkins, Board Member  
swilkins@barthsyndrome.org

#### Additional Contacts

Matthew J. Toth, PhD, Science Director  
mtoth@barthsyndrome.org

Lynda M. Sedefian  
Executive Administrative Assistant  
lsedefian@barthsyndrome.org

Leonard Steinberg, EA, CMC, Bookkeeper

Michael Hope, Webmaster  
mhope@barthsyndrome.org

Co-Chairs, Family Services:  
Shelia Mann, smann@barthsyndrome.org  
Chris Hope, chope@barthsyndrome.org

#### BSF Affiliates

The Barth Syndrome Trust  
(United Kingdom & Europe)

1 The Vikings

Romsey  
Hampshire  
S051 5RG

United Kingdom

Telephone: +44(0)1794 518785

E-mail: [info@barthsyndrome.org.uk](mailto:info@barthsyndrome.org.uk)

Website: [www.barthsyndrome.org.uk](http://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](mailto:inquiries@barthsyndrome.ca)

Website: [www.barthsyndrome.ca](http://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](mailto:jthorpe@barthsyndrome.org)

Website: [www.barthsyndrome.org/South\\_Africa.html](http://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 child(ren) may have Barth syndrome, you should consult your health care provider.

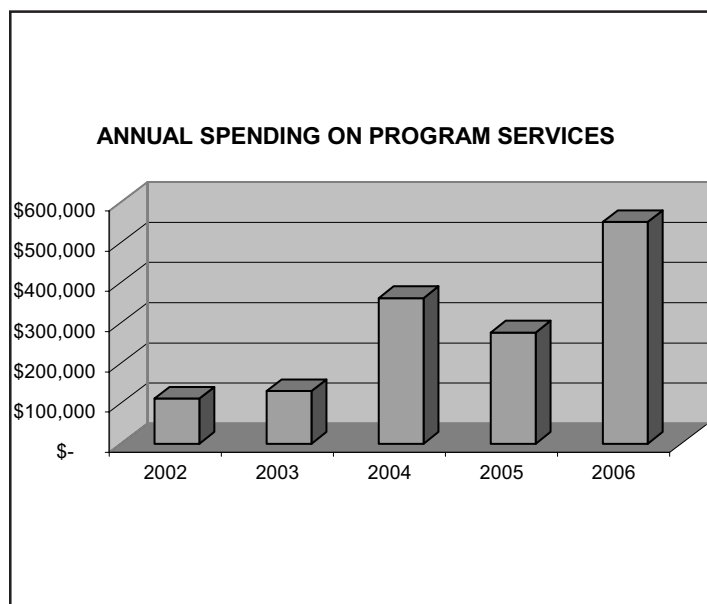
All submissions and correspondence regarding the newsletter should be directed to Lynda Sedefian, Newsletter Editor, at: [lsedefian@barthsyndrome.org](mailto:lsedefian@barthsyndrome.org).

# 2006: A year of great progress for BSF

**SCIENCE AND MEDICINE (cont'd from page 3)**

## **Medical Database and BioRepository**

The first substantive “deposits” were made into the Barth Syndrome Medical Database and BioRepository during the BSF Conference in July 2006. Dr. Carolyn Spencer is leading the first comprehensive effort to gather and document the many puzzling aspects of this complicated medical disorder. Dr. Spencer and the University of Florida have a multi-year grant from BSF to create the largest longitudinal database with matched biological samples (including DNA and cell lines) on Barth syndrome in the world. The existence of this database and its samples, along with the availability of research grants from BSF and larger institutions such as the National Institutes of Health (NIH), will attract new researchers to study Barth syndrome and accelerate research already under way. Dr. Spencer published a peer-reviewed article demonstrating the value these type of data can provide to inform physicians and healthcare providers about little known risks associated with BTHS. Her article addressed the prevalence of tachycardia in many Barth syndrome patients and demonstrated the benefit of implantable cardiac defibrillators to counter this risk – a finding that will undoubtedly save lives.



## **BSF's Research Grant Program**

In early 2006, BSF's research grant program awarded five grants totaling \$163,801 to researchers in Canada, Japan and the US, which brought the total number of grants awarded by BSF through 2006 to 19, representing over \$640,000. The details of these awards were described in Volume 6, Issue 1 of BSF's Newsletter. Earlier this year, BSF made additional awards of over \$309,200 to seven researchers in the US, Japan and the Netherlands, raising the total of grants awarded by BSF to just under \$1 Million. To date, BSF research grants have resulted in 17 articles that have been published in peer-reviewed medical journals, and stimulated additional, much larger research awards from institutions such as the National Institutes of Health (NIH), American Heart Association (AHA), and United Mitochondrial Disease Foundation (UMDF). Our strategy of awarding small (under \$40,000) “seed” grants to attract new researchers to the field of Barth research and to stimulate greater investment by larger funders is clearly working!

## **PHYSICIAN AWARENESS: STEVE KUGELMANN, PROGRAM LEAD**

As Dr. Colin Steward, a Pediatric Hematologist at Bristol Royal Hospital for Children in Bristol, England has written, “I vehemently believe that geneticists, cardiologists, haematologists, neurologists (muscle clinics), metabolic specialists and general paediatricians need to be aware of this disease. Only then can we reduce the risk of death through sepsis and heart failure, and offer carrier detection and antenatal diagnosis most effectively to affected families.” Physician awareness can mean survival for a newly born, affected boy. BSF continues to work to increase awareness through our newly re-designed website, the BSF Conference, participation in NIH meetings, the articles, presentations and posters written by our research grant award winners, and by setting up the BSF Booth at medical conferences. The booth is manned by BSF family volunteers, as well as members of our Scientific and Medical Advisory Board, when possible. We are often the only such booth in attendance and usually draw a fair amount of attention. In 2006 we attended 3 major medical conferences.

## **COMMUNICATIONS: LYNDA SEDEFIAN, PROGRAM LEAD**

We publish this Newsletter twice each year. It is one of our two broad scale communications vehicles (the other being our website). Our publications team strives to make it readable, engaging and informative for all of our various constituencies and it has been cited as a model by a number of other rare disorder groups.

In November we unveiled our newly redesigned website to provide broad, multi-language access to the single largest source of articles, reference materials, guidelines and information on Barth syndrome in the world. Visits to our website have been growing at a rate of over 38% per year since 2002, and this year exceeded 100,000, including for example, a record number of requests for information from students who selected Barth syndrome as the topic for their science report. To respond to this newfound interest in the disorder we have created a document which addresses the most common inquiries of these students. Pay us a visit at [www.barthsyndrome.org](http://www.barthsyndrome.org)!

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**OUR VOLUNTEERS: SHELLEY BOWEN, PRESIDENT OF BSF**

And now for the most amazing part! None of these accomplishments could have been achieved without the dedicated effort of our incredible volunteers. Today, BSF serves 107 affected families around the world, and countless others awaiting diagnosis or still agonizing over the unexplained symptoms threatening their children. Our families are separated by miles and kilometers, state and national boundaries, languages, medical systems and cultures. Barth Syndrome Foundation and its affiliates tie us all together via the internet, phone, this Newsletter, periodic meetings, and the BSF Conference. Our only employees are our President, Science Director and Executive Administrative Assistant. Many of our volunteers are family members who have a direct stake in the success of BSF's programs. But there are many others who have been inspired by the spirit of those families who are determined **never to give up** their search for a treatment and cure for Barth syndrome and a better life for their families, free from a disorder that revisits generation after generation. These are our volunteers, in the US, Canada, South Africa, Belgium, France, Italy, Portugal, the Netherlands, the UK, Australia and around the world, giving their time, optimism, energy and talent to a common vision – a future free from the threat of Barth syndrome. Our accomplishments are truly a reflection of teamwork at its finest. We are grateful to all who have donated their time and their service to our cause.

In October of 2006, 20 of these volunteers gathered together to plan the International Barth Syndrome Foundation's next five years. This is an extraordinary group, impatient for progress, driven to make life better for our children, and focused on the future. But on the first night Shelley Bowen and Lynda Sedefian rolled out a 20-foot scroll depicting a timeline of our accomplishments dating back to our first informal family meeting in Baltimore in 2000.

**YOU ARE ALL OUR VOLUNTEERS!**

Every family that donates blood, tissue, DNA and medical information in support of our medical database and biorepository; every scientist who works into the night in his or her lab striving to unlock the mysteries of the TAZ gene and Barth syndrome; every physician who takes time after work to search the thousands of pages of medical literature for an insight into the unique and sometimes frightening symptoms experienced by our children; every donor, touched by their personal connection to a Barth family and their faith in the ability of the International Barth Syndrome Foundation to make a real difference; and every family member and friend who raises money, visits medical conferences to increase awareness, writes an article for this Newsletter, helps build the BSF website, edits or translates a brochure, plans and organizes an outreach meeting, a clinic or the BSF Conference, reviews research grant proposals and insures that BSF's research investments are truly advancing our scientific understanding, or picks up the phone or sends an e-mail to another family looking for advice, counsel or simply a warm voice re-assuring them that they are not alone. Each and every one of you are our volunteers. We can never have enough of your time, your devotion and your dedication. And we can never thank you enough.

Our British families and friends will forgive us for borrowing a quote from Winston Churchill: **"Never... was so much owed by so many to so few"**.

<b>The Barth Syndrome Foundation, Inc. Statement of Activities For the Years Ended December 31</b>			
	<b>2006</b>	<b>2005</b>	
<b>Public Support and Revenue</b>			
Contributions	\$ 1,018,888	\$ 1,444,539	
Other	\$ 66,816	\$ 21,802	
<b>Total Support and Revenue</b>	<b>\$ 1,085,704</b>	<b>\$ 1,466,341</b>	
<b>Expenses</b>			
Program Services	\$ 550,838	\$ 276,544	
Management and General	\$ 98,875	\$ 90,655	
Fund Raising	\$ 4,115	\$ 14,900	
<b>Total Expenses</b>	<b>\$ 653,828</b>	<b>\$ 382,099</b>	
<b>Change in Net Assets</b>	<b>\$ 431,876</b>	<b>\$ 1,084,242</b>	
<b>Net Assets - Beginning of Period</b>	<b>\$ 1,897,278</b>	<b>\$ 813,036</b>	
<b>Net Assets - End of Period</b>	<b>\$ 2,329,154</b>	<b>\$ 1,897,278</b>	

**FINANCIAL GROWTH, TOO!**

By every financial measure, The Barth Syndrome Foundation had a good year in 2006. We raised over \$1 Million in contributions, doubled our investment in program services while keeping our administrative costs essentially flat, and increased Net Assets to over \$2.3 Million. But what does it all mean? Why should we all be very proud of what we are all creating together here? Let's take it one step at a time.

**Administrative Costs**

Charity watchdogs (including the IRS and the 13 states with which BSF registers each year) rightly focus on the percentage of funds raised that go toward non-program (i.e. Management and General plus Fundraising) expenses. For 2007, this figure was 10.1%, up from the prior year of 7.3% but well within acceptable standards. BSF continues to run a tight ship!

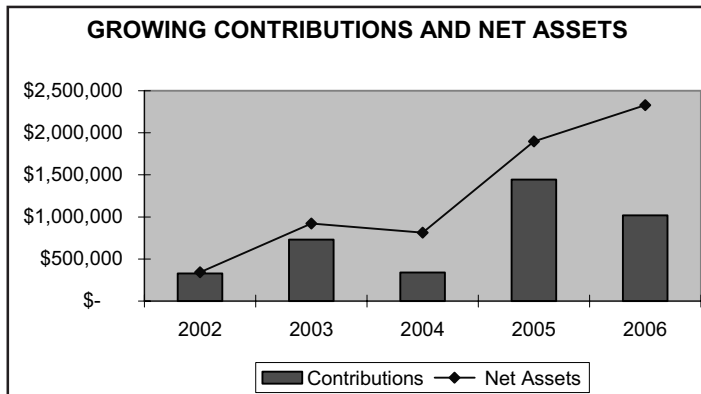
(Cont'd on page 6)

# 2006: A year of great progress for BSF

(Cont'd from page 5)

## GROWING CONTRIBUTIONS AND FUND BALANCES

*Near Record Contributions in Fundraising: Leslie Buddemeyer, Program Lead*



We raised \$1,018,888 from over 660 contributors in 2006 – second only to 2005 when we kicked off our long-term Science and Medicine fundraising effort and received a number of particularly large donations. In 2006, 18 donors gave BSF more than \$10,000 each – they are our “Super” Stars on our *Power of Kindness* section within this Newsletter (pgs. 8 thru 10).

In every case, donations came to BSF because someone in a family affected by Barth syndrome, or a friend, *asked* for a donation.

- Tom and Laurie Monahan and their friends in Brockton, MA continue to be strong BSF supporters with multiple fundraisers in the past year!
- Gary Rodbell, BSF’s Ironman, and his friends used their interest in triathlons to inspire their friends to contribute over \$310,000 to BSF last year!
- John and Liz Higgins invited their friends to the Third Annual Bowling for Barth fundraiser, had a great time and raised money for BSF at the same time. Mark your calendars, the Fourth Barth Bowlathon is scheduled for October 13, 2007!
- Joyce Lochner’s friend Nina Rivers made a beautiful Barth Quilt and Joyce and the Barth kids auctioned it off at the BSF Conference in July.
- Randy Buddemeyer and his colleagues at C.B. Richard Ellis in Florida sponsored a golf tournament that raised \$20,000 for BSF from their friends and business associates. They asked 144 of their friends and business associates to enjoy a great day of golf and help BSF... and every tee time was taken.
- Scott Oldewage continues to work with his employer Ed Pace and Lake City Trucks in Utah to raise funds through employee giving. Ed Pace then matches everything his employees give to BSF and adds his own generous contribution each month. This program and others like it are becoming major sources of contributions for BSF!
- Sue Wilkins and her mother, Paula Varner, continued to write to and personally ask family friends to support the Woody Varner Science and Medicine Fund, named after Sue’s dad and John’s grandfather. They raised an additional \$57,000 for BSF in 2006.
- Eighty-six members of the Congregation of St. John’s Church in Larchmont responded to the Church’s Easter Appeal and contributed over \$53,000 to BSF. The McCurdy family was the link between the St. John’s and Barth communities and Steve McCurdy spoke from the pulpit about the strength and support that the family drew from each of these groups.
- The McCurdys have also written an annual letter to their friends, family, school classmates and business colleagues to tell them about the progress BSF was making and the importance of BSF to their son Will’s life. For seven years, their friends have remained faithful and increasing supporters of BSF – an expression of love and support for which the family is forever grateful.

It’s increasingly clear. The Barth families and their friends have a powerful story to tell and everyone benefits whenever the story is told. People want to help. And in 2006 they did so by contributing over \$1 Million to BSF. Thank you all!

We would also like to thank the volunteers in our fundraising committee, Laurie and Tom Monahan, John Higgins, Scott and Casie Oldewage.

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(Cont'd from page 6)

<b>The Barth Syndrome Foundation, Inc.</b>			
<b>Statement of Financial Position</b>			
<b>At December 31</b>			
	<b>2006</b>	<b>2005</b>	
<b>Assets</b>			
Current Assets	\$ 2,460,331	\$	2,053,960
Other Assets		\$	-
<b>Total Assets</b>	<b>\$ 2,460,331</b>	<b>\$</b>	<b>2,053,960</b>
<b>Liabilities</b>			
	\$ 131,177	\$	156,682
<b>Net Assets</b>			
Unrestricted	\$ 1,939,939	\$	1,870,488
Temporarily Restricted	\$ 389,215	\$	26,790
<b>Total Net Assets</b>	<b>\$ 2,329,154</b>	<b>\$</b>	<b>1,897,278</b>
<b>Total Liabilities and Net Assets</b>	<b>\$ 2,460,331</b>	<b>\$</b>	<b>2,053,960</b>

### GROWING NET ASSETS

Another area that properly draws attention is our growing Net Asset position. Our Net Assets grow when we do not spend all of the money we raise and fall when we spend more than we raise. BSF has been fortunate to have had a number of very successful fundraising efforts in past years and has been able to grow our programs as a result. However, the Board is mindful that BSF is still a small foundation, supported almost entirely by volunteers. Fundraising can vary significantly from year to year.

For this reason, as you may recall from previous newsletters, BSF's Board launched a multi-year Science and Medicine fundraising campaign with the objective of pre-funding our Science and Medicine programs through 2015. These programs (including our annual Research Grant program, the Barth Syndrome Conference, the Barth Syndrome Medical Database and BioRepository, our physician awareness

program and a series of scientific and medical meetings sponsored by BSF) are on-going, and we do not want them to slow or stop. Momentum is an important ally and we want it working for us, not against us! BSF's Net Assets are "insurance" against the loss of scientific and medical momentum in future years should annual fundraising fall short or extraordinary opportunities to invest in new advances come up, and we hope that they will continue to grow in years to come.

In terms of budgeted Science and Medicine Program costs, our current Net Assets should just about cover these programs for the next four years. We still have \$3.6 Million more to raise for Science and Medicine if we are to achieve our goal. In the meantime, these funds are invested and should produce approximately \$115,000 in interest earned in 2007, which can be used to offset our administrative costs.



## WITH DEEPEST APPRECIATION

At long last, it can be acknowledged. BSF has recently been given permission to publicly identify our devoted "Anonymous Donor" who has supported BSF since our very first year. The friend and supporter is The Annenberg Foundation in Pennsylvania. The Annenberg Foundation has a long history of identifying and supporting causes that truly add to the fabric of American society. As defined by Ambassador and Mrs. Walter Annenberg, the Foundation's focus includes: Education and Youth Development; Arts, Culture and Humanities; Civic, Community and the Environment; and Health and Human Services. In line with Ambassador Annenberg's lifetime interests, the Foundation's Mission is "Advancing public well-being through improved communication." You can learn more about the Annenberg Foundation at [www.annenbergfoundation.com](http://www.annenbergfoundation.com).

BSF came to the attention of Mrs. Annenberg and the Annenberg Foundation through her strong personal connection with Mary Baffa and the Baffa Family. Her concern for Kevin Baffa's well-being extended generously to the Barth Syndrome Foundation, and The Annenberg Foundation has been a key donor to BSF in every year since our founding. Support from such an established and well respected donor has inspired us all to set very challenging goals and then given us the confidence to achieve or exceed them. We know we have a marathon in front of us, but the race is a little bit easier when we have such a strong and caring running mate. We are all eternally grateful and look forward to the support of the Annenberg Family and the Annenberg Foundation for many years to come.



# Power of kindness

(Contributions donated since January 2006)

## WOODY VARNER FUND

### Stars (\$5,000+)

Andersen, Harold/Marian  
Daugherty, Robert  
Dillon Foundation  
Mawby, Mr./Mrs. Russ  
Richard & Virginia Voell Family Fund  
Suzanne and Walter Scott Foundation  
Wilkins, Dr. Mike/Sue  
Yanney, Michael/Gail

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(Cont'd on page 10)

# Power of kindness

(Contributions donated since January 2006)

## Time and Advice (cont'd)

Day, Dr. Jane  
De Decker, Dr. Rik  
Develle, B.J.  
DiMauro, Dr. Salvatore  
Doherty-Bigara, Jérôme  
Dowhan, Dr. William  
Duggan, Adela  
Dunbar, Terry  
Drinkwater, Paul  
Duncan, Peter CPA  
Easterbrook, Isabel/Ralph  
Elwood, Lynn  
Elwood, Rick  
Epstein, Paul  
Fabian, Joan  
Fioole, Johan  
Final Focus Productions (Steve Kownacki)  
Fortier, Jodie  
Foxon, Kim  
Frost, Tracy  
Gagnon, Lou Ann  
Galbraith, Lois  
Ganz, Doug  
Gaudin, Herve  
Geigle, Dr. Paula  
Gemmell, David  
Gerszberg, Rich  
Geva, Dr. Tal  
Gill, Simon at Keens Shay Keens  
Gilmour, Lisa  
Gilmour, Lorraine  
Gonik, Renato  
Gonzalez, Dr. Iris  
Gordon, Karen  
Gravitt, Carolyn  
Greenberg, Dr. Miriam  
Groft, Dr. Stephen  
Guerin, Olivier  
Guimarey, Ilda  
Haines, Dr. Thomas  
Hall, Colin/Midge  
Hall, Dacey  
Hall, Teresa  
Hancock, Lynn  
Harford, Greg  
Harford, Larry  
Harley, Elma Rhea  
Harvath, Dr. Liana  
Hawkins, Lawton  
Hatch, Dr. Grant  
Hauff, Kristin  
He, Dr. Quan  
Heal, Elisabeth  
Henderson, Camilla  
Hennekam, Prof. Raoul  
Hennessey, Mike  
Henry, Anne  
Henry, Dr. Susan  
Hicks, Sarah  
Hintze, Audrey  
Hobbs, Mark  
Holly, Keli  
Holmes, Pam  
Hone, Susan  
Hope, Christiane  
Hope, Michael  
Houtkooper, Dr. Rieket  
Jensen, Joanne  
Jensen, Dr. Robert  
Johnson, Benjamin and Family  
Johnson, Linda  
Johnson, Kristen  
Juico, Eileen  
Kacinski, Debbie

Kainer, Daryl  
Karp, Matt/Wendy  
Kearns, Richard/Cathy  
Kennedy, Yongyong  
Kelley, Dr. Richard  
Kern, Rebecca  
King, Bobbie  
King, Lynn  
Knauer, Bill  
Kobayashi, Dr. Toshihide  
Kovacs, Dr. Adrienne  
Kroon, Dr. Anton de  
Kropp, Susan  
Kugelmann, Dave  
Kugelmann, Irene  
Kugelmann, Jan  
Kugelmann, Matt  
Kugelmann, Steve  
Kuijpers, Dr. Brett  
Kulik, Dr. Willem  
Lamoia, Michelle  
Lane, Anna  
Langendonck, Veerle van  
Lauring, Dr. Brett  
Lawson, Lee Ann  
Layton, Alanna  
Leça, Dr. Ana  
Leckford Estate, Stockbridge  
Lee, Dr. Ray  
Lemette, Isabelle  
Lever, Beverly  
Lever, Jeff  
Levin, Dr. Gail  
Lewin, Dr. Tal  
Lewis, Julie  
Lo, Dr. Cecilia  
Lobe, Steve/Hilary  
Lochner, Joyce  
Loo, Albert/Jo van  
Lowe, Jodi  
Lynn, John  
Lyll, Doug  
Madgett, Roberts, Marlowe, Jackson & Associates  
Mah, Dr. Cathryn  
Maisenbacher, Melissa  
Mancino, Angelo  
Mann, Shelia  
Manton, Annick  
Manton, Rob  
Manton, Greg  
Manton, Joan  
Margossian, Renee  
Martin, Joy  
Martin, Dr. Rob  
Martins, Raquel  
Maruno, Yuriko  
Mason, Taylor  
Matthias, Linda  
Maxfield, Dr. Frederick  
Mazzocco, Dr. Michele  
McConaughy, Bobbi  
McConaughy, Jim/Bev  
McCurdy, Kate  
McCurdy, Eliza  
McCurdy, Steve  
McCurdy, William  
Meisenbacher, Melissa  
Merlo, Lisa  
Mileykovskaya, Dr. Eugenia  
Miller, Cheryl  
Mitchell, Jim  
Mock, Kim  
Monahan, Laurie  
Monahan, Tom  
Montenero, Theresa

Moore, Lorna  
Moore, Nigel  
Moore, Dr. R. Blaine  
Morava, Eva  
Morgenstern, Marc  
Morris, Dr. Andrew  
Morrison, Cecilia  
Morris, Les  
Murphy, Tony  
Nackashi, Dr. John  
Nash, Alan/Lesley  
Newbury-Ecob, Dr. Ruth  
Nicoll, Rev. Tom  
Nuckolls, Dr. Glenn  
Oldewage, Casie  
Oldewage, Scott  
Olson, Dean/Tina/Brandy  
Olson, Adam  
Olson, Maria/Tom  
Olson, Sharon/Richard  
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Osno, Susan  
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Parrish, Dr. Catherine  
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Pruett, Debbie  
Queenan, Jeri Eckhart  
Radar, Stephanie  
Radosta, Lori  
Read, Yvonne & Playleaders  
Redfeare, Sharon  
Reedy Creek Emergency Services  
Reece, Bryce  
Rehling, Dr. Peter  
Reiss, Dr. John  
Ren, Dr. Mindong  
Reppen, Heather  
Riet, Hans van der  
Rinaldo, Dr. Piero  
Ritter, Cathy  
Rivers, Nina  
Rodbell, Gary/Collette/Julia  
Rosen, Amy  
Rosenshine, Jon/Draddy, Mary Pat  
Roubos, Mr./Mrs.  
Ryan, Dr. Robert  
Satula, Deborah  
Schantzen, Sandy  
Schlame, Dr. Michael  
Schroeder, Raghad  
Schroeder, Wallace  
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Segui, Damaris  
Shaddy, Dr. Robert  
Shenkman, Dr. Elizabeth  
Sherbany, Dr. Ariel  
Sherman, Cathy  
Sheppard, Jamie  
Sherwood, Dr. Geoff  
Shirley, Alan/Denise  
Shum, Bill/Ginny

Smith, Deborah  
Smithson, Sarah  
Smolski, Ed  
Snyder, Floyd  
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Storch, Jill  
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Walker, Gena  
Wanders, Dr. Ronald  
Ward, Barry  
Ward, Anne  
Watson, Karin  
White, Marty  
Wicker, Judy

Wiederspan, Jess  
Wierenga, Dr. Klaas  
Wiggins, Mark  
Wilkins, John  
Wilkins, Dr. Michael  
Wilkins, Sue  
Wilks, Carol  
Williams, Kathy  
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Wise, Lisa  
Wood, Robin  
Wroe, Martha  
Xu, Dr. Yang  
Zurbrick, Patty

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Thank you for your patience as we continue to update our records!



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# A global roadmap for Barth syndrome research

(Cont'd from Cover)

## **Pathogenetic mechanism and genetic suppressors of Barth syndrome**

Mindong Ren, PhD, of New York University, will build on his previous success of developing a fruit fly model of Barth syndrome. At the July 2006 BSF Scientific/Medical and Family Conference in Orlando, his presentation of a fruit fly model of Barth syndrome showed some very useful qualities that a

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*“This approach has the potential to discover many different ways to overcome or correct the tafazzin dysfunction, which we hope will lead toward finding a specific treatment for Barth syndrome.”*

---

scientist can use to tease out how the protein tafazzin interacts with other proteins. In a powerful genetic system like the fruit fly, Mindong will be testing what other mutants can reverse the qualities he observes with his tafazzin deletion flies. This is known as suppressor analysis in genetics, and it can show connections among proteins/genes which cannot be seen in any other way. This approach has the potential to

## **Identification of the proteins interacting with tafazzin and resolution of the consequences of the deficiency of cardiolipin at the protein level**

Frédéric Vaz, PhD, of the Amsterdam Medical Center, will be using sophisticated molecular techniques to discover what proteins physically interact with the tafazzin protein. By placing a mark or tag on the tafazzin protein, Fred will isolate those proteins that are able to interact with or to stick to tafazzin. By identifying these “sticking/interacting proteins” we may discover what other genes may alter or may even correct a tafazzin dysfunction. These experiments are analogous to what Mindong Ren is doing with his genetic fruit fly model, but Fred will be working with protein instead of DNA which requires different tools and techniques.

## **The role of phosphatidylglycerol in activating Protein Kinase C mediated signaling**

Miriam Greenberg, PhD, of Wayne State University, will be using another powerful genetic system, yeast, to probe the biochemical connections of genes that synthesize the lipid cardiolipin, the biomolecule most affected in Barth syndrome. She will be looking into how proteins involved in the cardiolipin biosynthetic pathway, such as mutations in the tafazzin gene, turn genes on or off. The results should lead us to understand how differences in lipid molecules like cardiolipin can give rise to the different symptoms suffered by Barth patients.

## **Regulation of cardiolipin remodeling in the heart studied using a rat model of heart failure**

A new investigator to our BSF family, Genevieve Sparagna, PhD, of the University of Colorado, will be looking into how the heart is remodeled after suffering from cardiac disease. While studying a famous rat model of heart disease (the spontaneous hypertensive heart failure model—SHHF) Genevieve found that tafazzin protein levels were significantly altered as the heart repaired itself in a process called remodeling. How tafazzin contributes to this process of heart remodeling is not understood, but its relevance to the heart problems suffered by the boys is interesting. She will be using this same rat model of heart disease to test whether certain chemical compounds known to alter the remodeling process can prevent or reverse these phenomena.

## **Development of BTHS screening using bloodspots and HPLC tandem mass spectrometry**

Willem Kulik, PhD, also of the Amsterdam Medical Center, will be critically testing the cardiolipin bloodspot test he and his co-workers have been developing. This will provide a second test to determine a diagnosis of Barth syndrome, like the genetic test. This type of test may also be very useful as a way to objectively monitor the severity of Barth syndrome. We know that the lipid cardiolipin is the major alteration observed in Barth syndrome patients, but we do not know if this correlates with disease severity. Willem’s work may provide proof of this correlation, which if true, would also be a way to objectively measure whether a drug treatment is actually working.

## **Early indices of learning difficulties in young boys with Barth syndrome**

On the clinical side of the research, Michele Mazzocco, PhD, of Johns Hopkins, will be testing the mental abilities of preschool age boys. Because of the hardships endured by Barth boys, schooling and intellectual development of Barth boys may be negatively affected by fatigue and not caused directly by Barth syndrome. This fatigue may influence any determination of mental abilities resulting from testing. To more rigorously answer the question of whether Barth boys have mathematical/visual-spatial problems, Michelle will be testing several preschool age boys.

## **Cardiac and skeletal muscle in Barth syndrome: Evaluation of functional capacity and energy metabolism**

Carolyn Spencer, MD, of the University of Florida, will be monitoring blood levels of substances, cardiovascular parameters, and muscle biochemistry of certain Barth syndrome patients. By comparing the measurements to those of unaffected boys, Carolyn should be able to provide some objective criteria to monitor Barth patients as they become older. This information will also be useful in determining if a treatment is beneficial, by comparison with the normal changes in these parameters in Barth syndrome patients.

(Cont'd on page 12)

# A global roadmap for Barth syndrome research

(Cont'd from page 11)

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*“This is a great sign of progress in our mission to find a treatment or a cure for Barth syndrome.*

*I like to characterize investigational science as similar to putting together a picture puzzle of a thousand pieces without using the box as a guide. As in the picture puzzle, the more pieces we can fit together, the more visible the picture becomes.”*

---

## Organization of cardiolipin in Barth syndrome

Toshihide Kobayashi, PhD, of the RIKEN (The Institute of Physical and Chemical Research) in Japan, will continue with his studies of the lipid cardiolipin. We know that the unique lipid molecule cardiolipin is altered and decreased in patients with Barth syndrome. Dr. Kobayashi is trying to understand how the dysfunction of this lipid is associated with many of the symptoms that Barth patients suffer from. He will employ techniques that measure the physical properties of cardiolipin-containing membranes such as X-ray scattering, calorimetry, and scanning tunneling microscopy. In addition, Dr. Kobayashi will be generating tools (antibodies) that can specifically identify the cardiolipin molecules in the membrane. We anticipate that by knowing how cardiolipin alters membranes, we may better understand how the Barth patients are affected.

## SUMMARY

The current crop of awards is composed of many different areas of research. Many of these build on the progress and the data derived from earlier awards. This is a great sign of progress in our mission to find a treatment or a cure for Barth syndrome. I like to characterize investigational science as similar to putting together a picture puzzle of a thousand pieces without using the box as a guide. As in the picture puzzle, the more pieces we can fit together, the more visible the picture becomes. In addition, as more pieces are correctly placed, the easier it is to find out where any particular unplaced piece goes. When we have a more complete picture of Barth syndrome, we should be in a strong position to realize our mission.



## TIMELINE

### BARTH SYNDROME MEDICAL DATABASE AND BIOREPOSITORY

#### July 2006

- Collection of medical records, patient demographics, DNA and lymphoblast cell lines
- 37 enrolled

#### June 2007

- Website open to public: [www.peds.ufl.edu/barthsyndromeregistry/](http://www.peds.ufl.edu/barthsyndromeregistry/)

#### July 2007

- Completion of Intake Forms

#### September 2007

- Advertise for enrollment
- Abstraction of data from medical records
- Merged data from Cardiac Research Study
- Accept outside requests for data and biological samples

#### Medical Database and Biorepository Advisory Board

**Carolyn Spencer, MD (Principal Investigator), Division of Pediatric Cardiology, University of Florida College of Medicine, Gainesville, Florida**

Melissa Maisenbacher, MS, CGC (Medical Data Abstraction, *ex-officio*), University of Florida College of Medicine, Gainesville, Florida

Deepa Ranka, MS (IT and Statistical Research Analyst, *ex-officio*), Department of Epidemiology and Health Policy Research, University of Florida College of Medicine, Gainesville, Florida

Richard I. Kelley, MD, PhD, Division of Metabolism, Kennedy Krieger Institute, Johns Hopkins University, Baltimore, Maryland

Matthew J. Toth, PhD, Barth Syndrome Foundation Science Director, Iselin, New Jersey

Gerald Cox, MD, PhD, Senior Medical Director, Genzyme Corporation, Cambridge, Massachusetts

Michael Schlame, MD, Department of Anesthesiology, New York University School of Medicine, New York, New York

Colin G. Steward, FRCP, FRCPCH, PhD, Pediatric Hematology, Bristol Royal Hospital for Children, Bristol, England

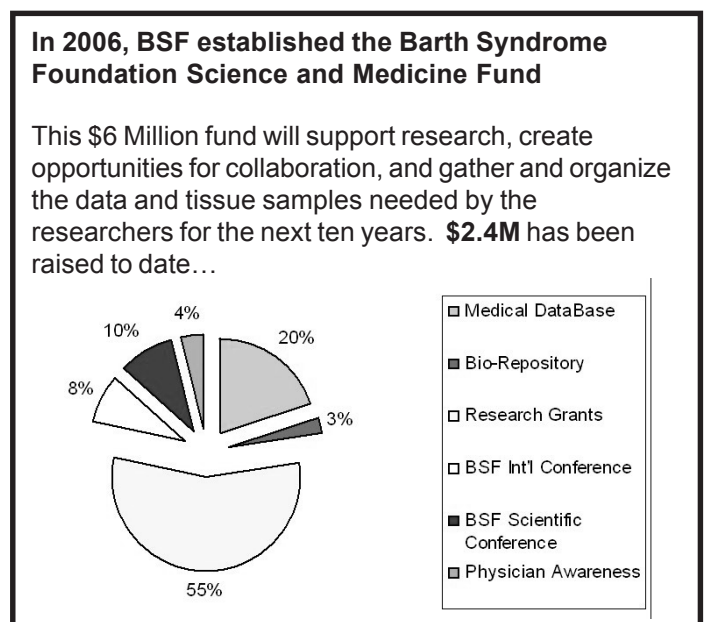
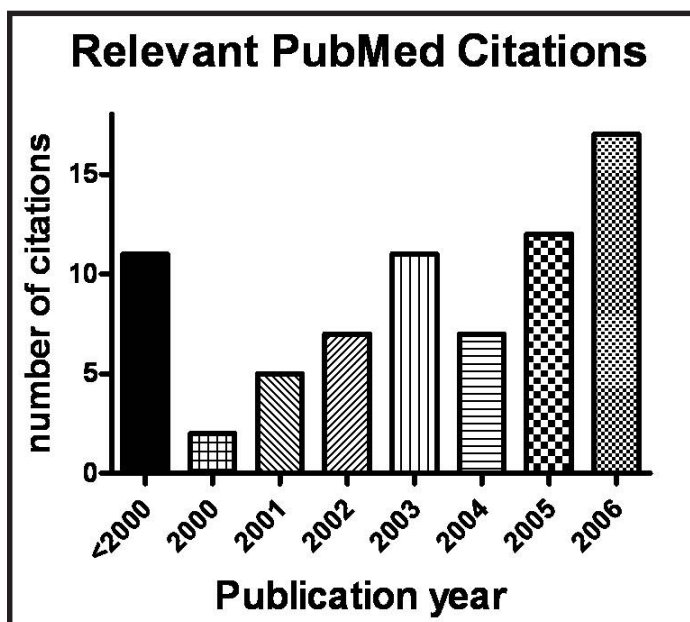
# Barth syndrome bibliography expands

## A compelling increase as a result of BSF funding

There is a statistically significant increase in Barth syndrome related peer-reviewed journal articles published recently, which is due in part to BSF's successful research grant program. To date, there have been 17 journal articles published as a direct result of BSF funding.

Listed below are the latest articles added to BSF's library:

1. Sparagna GC, Chicco AJ, Murphy RC, Bristow MR, Johnson CA, Rees ML, Maxey ML, McCune SA, Moore RL. **Loss of cardiac tetralinoleoyl cardiolipin in human and experimental heart failure.** J Lipid Res. 2007 Apr 10; [Epub ahead of print].
2. Mangat J, Lunnon-Wood T, Rees P, Elliott M, Burch M. **Successful cardiac transplantation in Barth syndrome - single-centre experience of four patients.** Pediatr Transplant 2007 May;11(3):327-331.
3. Marziliano N, Mannarino S, Nespoli L, Diegoli M, Pasotti M, Malattia C, Grasso M, Pilotto A, Porcu E, Raisaro A, Raineri C, Dore R, Maggio PP, Brega A, Arbustini E. **Barth syndrome associated with compound hemizygosity and heterozygosity of the TAZ and LDB3 genes.** Am J Med Genet A. 2007 Mar 29; [Epub ahead of print].
4. Crawford J. **A 3D view of Barth syndrome mitochondria.** Lab Invest. (2007), 1-2.
5. Mazzocco MM, Henry AE, Kelly RI. **Barth syndrome is associated with a cognitive phenotype.** J Dev Behav Pediatr. 2007 Feb;28(1):22-30.
6. Kirwin SM, Vinette KM, Schwartz SB, Funanage VL, Gonzalez IL. **Multiple transmissions of Barth syndrome through an oocyte donor with a de novo TAZ mutation.** Fertil Steril. 2007 Jan 19; [Epub ahead of print].
7. Day JA, Spencer CT, Byrne B. **Grip Strength in a Population of Boys with Barth syndrome.** Pediatric Physical Therapy Vol. 18(1)88-89 Spring 2006.
8. Szulik M, Lenarczyk A, Rycaj J, Bialkowski J, Dziubek B, Kalarus Z, Kukulski T. **Isolated non-compactation of the left ventricular myocardium in a neonate - a case report.** Kardiol Pol. 2006 Dec;64(12):1422-5.
9. Alessandri C, Sorice M, Bombardieri M, Conigliaro P, Longo A, Garofalo T, Manganelli V, Conti F, Esposti ED, Valesini G. **Anti-phospholipid reactivity against cardiolipin metabolites occurring during endothelial cell apoptosis.** Arthritis Res Ther. 2006 Dec 6;8(6):R180 [Epub ahead of print].
10. Brady AN, Shehata BM, Fernhoff PM. **X-linked fetal cardiomyopathy caused by a novel mutation in the TAZ gene.** Prenat Diagn. 2006 May;26(5):462-5.



## What is Barth syndrome?

**Barth syndrome (BTHS)** 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:** Abnormal growth pattern, similar to, but more severe than, constitutional growth delay. Most boys with Barth syndrome are below average in weight and height, often substantially so, until the late teenage years.

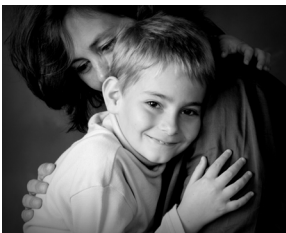
**Early and accurate diagnosis is key to survival for Barth syndrome boys.** Historically, boys died of heart failure or infection by 3 years of age, but today, with improved diagnosis, treatment, and management, the survival rate and future of these boys is much brighter.



*Edmond and William  
Georgia*



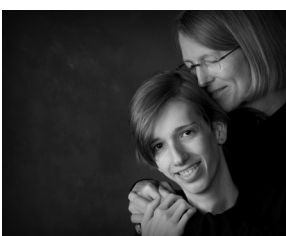
*Shelia and Jeremiah  
Maryland*



*Allison and Colin  
South Africa*



*Rod, Annie, and Matt  
Australia*



*Kate and Will  
New York*

## Who is BSF? A Committed Group of Families

## 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. Families of children with a chronic illness 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.

# Focus on Family

## Signs, symptoms, and obtaining a diagnosis

By Julie Floyd, Georgia

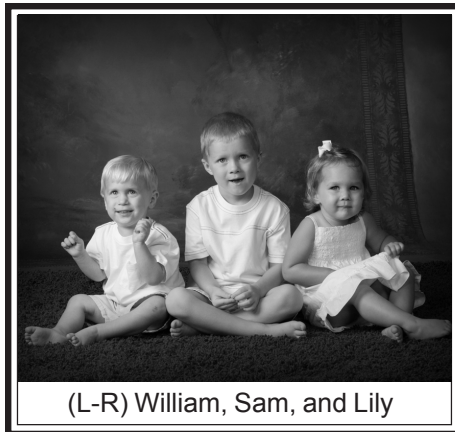
**W**e are the Floyds from Dacula, Georgia. Ed and I have a wonderful 5 year-old son Sam and precious twins, William and Lily, who have just celebrated their 3rd birthday. There is NEVER a dull moment in our home.

Being pregnant with twins is a wild ride for sure. There were many days as I approached “D” day, that I truly thought that I would spontaneously combust as I caught my then 2-year old son, Sam, as he came down the slide at the park. However, as you can probably figure out, thankfully my worst fears were never realized!

My obstetrician became concerned after approximately 22 weeks into my pregnancy with William and Lily. Twin A (William) was not growing at an acceptable rate. I was having ultrasounds every 14 days to monitor growth to see if our babies would need to be delivered early in order to save Twin A (William). We were counting days and praying each day for both babies to be born healthy. Well, that turned out in hindsight to be a joke – at 37 weeks the doctors had to force the two little monsters out!

My first words when William was delivered were, “Is he healthy? Is he all right?” We were assured that he was just fine and that obviously all that worrying during my pregnancy had been for nothing. He weighed in at 5 pounds 6 ounces and was adorable.

Fast forward, if you will, to William’s 6 month check-up with the pediatrician. You see, William was not performing the “tricks” that Lily was (like sitting up), and he was not anywhere on a growth chart. So, we began physical therapy, and he slowly mastered the skills that he needed. It must just be an isolated case of hypotonia, right?



We then visited a neurologist who also felt that there probably was no “bigger picture” for William – we just needed to continue physical therapy and at that point we also added occupational therapy to help his fine motor skills which also seemed delayed.

Fast forward once again to William’s 15 month check-up – the first with a new pediatrician that I had chosen just because I felt we needed a change. It was one in a chain of several appointments over the next few months that I will never forget. The doctor said (after listening for what seemed like FOREVER to his heart), “Now, who is William’s cardiologist?” My heart sank. “His what?” I said. I still thank God each day for sending us to this new pediatrician.

Within a matter of a few days, we were in the office of a wonderful cardiologist. We were told that William was on the verge of heart failure. He was immediately started on meds, and all kinds of scary terms and procedures were thrown out to us. I truly felt broken at that point. A few days after this appointment, William’s cardiologist called us and said that after giving much thought to William’s situation, he recommended that we see a geneticist. He believed that all of these random pieces did in fact fit together into a bigger picture for William. I know what you must be thinking. How could we not have figured this out earlier?

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*“...I believe that all things happen for a reason.*

*William is ours and the people within the Barth Syndrome Foundation are ours for a reason.*

*For that, we continue to be so thankful.”*

---

After initial genetic testing and what seemed like 78 pages of medical history of anyone who we had ever even considered to be family, we tested for Barth syndrome. Our geneticist directed me to the Barth Syndrome Foundation website for information on this disorder. Of course, being the proactive mother that I am, I immediately went to the website and just about as quickly clicked on the “x” in the upper right hand corner. Potentially fatal? Fatigue? Abnormal Growth? Heart Failure? Neutropenia? I was not ready for this!

I will never forget finally getting the courage to submit an email on the website. Within 12 hours, I had talked with Shelley Bowen, Sheila Mann, and Sue Wilkins. Wow! I instantly felt a sense of family and had found people who could truly understand our hopes and fears for our precious William.

You see, I believe that all things happen for a reason. William is ours and the people within the Barth Syndrome Foundation are ours for a reason. For that, we continue to be so thankful. Our family is enormously blessed. We have three wonderful children that we love beyond anything. William is now eating like a horse (or 2), has achieved the status of being on the growth chart, is as smart as they come, and absolutely loves playing with his brother, sister, and friends. I am reminded each day to enjoy every moment with my children – I don’t know what tomorrow holds but I sure can have lots of fun today!



# Celebrating a milestone

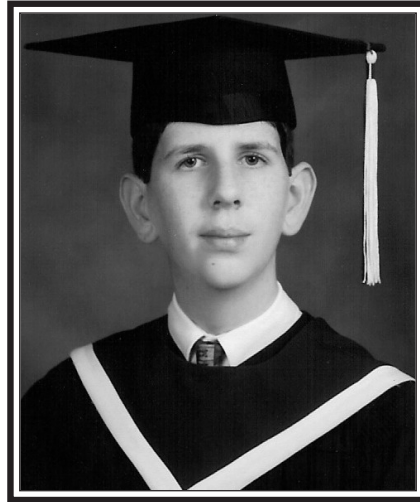
## Barth men are making huge strides

By Alanna Layton, Sister of Barth Individual

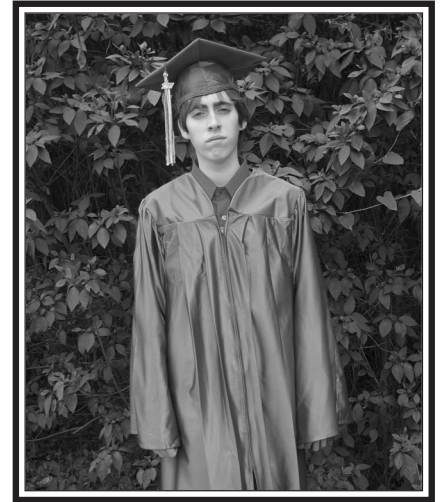
High school graduation is a major milestone in any person's life and is a time to be celebrated. Many of our young men with Barth syndrome have encountered unique challenges during their school years, unparalleled by their peers. I am honored to have been asked to write about the triumphs of three extraordinary young men who graduated high school during the summer of 2007, and I believe it is no understatement to say that we are all incredibly proud of their achievements in school - and in life!



**Andrew Buddemeyer** graduated from H.B. Plant High School in Tampa, Florida on May 18. *"My high school experience was good. I really enjoyed it,"* Andrew said. His school was willing to work with him whenever possible, including limited physical education and having an extra set of books for his classes. Even working three days a week as a cashier after school, Andrew was able to maintain 4.0 GPA throughout high school and graduate on time for his age. He was also a member of several different student honor societies. *"I really believe school is important – it helps to prepare you for the real world,"* he said. He will attend the Honors College at Florida State University in the fall. Because of his course work in Accelerated Placement (AP) classes, Andrew will enter college as a sophomore. *"I am looking forward to attending Florida State in the fall and moving on to this next stage in life!"*



**Kevin Baffa** graduated from Bishop Shanahan High School in Downingtown, Pennsylvania on June 1. *"I really enjoyed my high school experience. I made a lot of great friends, friends I didn't even know I had,"* he said. For Kevin, his biggest challenge was trying to keep his energy up and taking time to slow down. *"I would take one day off a week to rest and get my energy up. That helped a lot."* He was an honors student through most of his high school years and is taking the fall off to explore his vocational and educational options. He does want to attend college in the near future. *"To the younger guys who are struggling with school, I would tell them that school is hard but you can adapt. People are willing to work with you."*



**Michael Bowen, Jr.** graduated from Taylor County High School in Perry, Florida on May 18. *"My high school experience was unique because I was home-schooled most of the time. Going back to a regular classroom setting this last year was different because of all the people there,"* he said. He was awarded a scholarship from North Florida Community College for his academic accomplishments. In addition he has received a scholarship from the State Vocational Rehabilitation Block Grant until he completes his college education. *"I am very excited about finishing high school and celebrating with my family. I am also very excited about starting college in the fall!"*

As Michael's sister, I witnessed his challenges. As a family, we were there to support Michael, just as the other families supported their boys. While we all share in the joy of their triumphs, these three young men walked the path. Congratulations are certainly in order for their hard work, determination, accomplishments, and a job well done!





# Sibling spotlight

## Featuring friends from U.S. and U.K.

By Alanna Layton, Sister of Barth Individual

**B**elow are the profiles of three of our fantastic BSF siblings. It is wonderful to see siblings of those affected with Barth syndrome form new friendships among our community of “Barth Families.” We stongly believe that these relationships are so meaningful and will be everlasting!

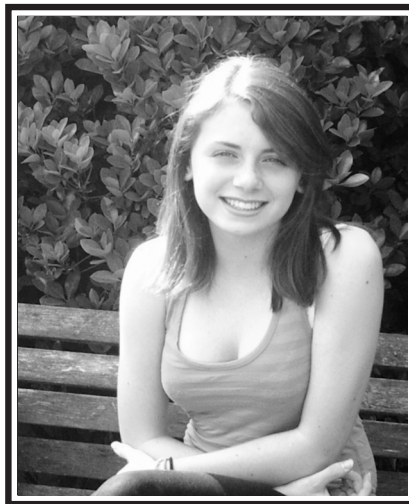


**Name:** Nicholas “Nick”  
**Age:** 11  
**Where are you from:** The United Kingdom  
**Affected sibling:** Alexander “Alex”

**What is your favorite thing to do with your brother:** Playing football together and playing PlayStation together

**If you met someone who just found out that their brother had Barth syndrome, what would you say to them that might make them feel better:** Barth syndrome affects different boys in different ways. It doesn't stop them from being able to do things; they just get tired a lot quicker and need to rest a lot more.

**What does BSF mean to you? Why is it important to you:** I know that BSF is doing all it can to find a cure for my brother's illness. I also now know that there are a lot more families out there going through the same as my family. We are not alone.



**Name:** Eleanor “Ellie”  
**Age:** 12 (almost 13!)  
**Where are you from:** The United Kingdom  
**Affected sibling:** William

**What is your favorite thing to do with your brother:** Spend time on the PlayStation game “Crash Team Racing” together.

**If you met someone who just found out that their brother had Barth syndrome, what would you say to them that might make them feel better:** That it is no different from having a brother without it, except he may not be able to do some things you'd like to do with him because he gets so tired too easily.

**What does BSF mean to you? Why is it important to you:** I feel that I am very connected with it and it makes me feel very involved with it.



**Name:** Jamie  
**Age:** 16 (17 as of June)  
**Where are you from:** West Chester, Pennsylvania  
**Affected sibling:** Kevin (18)

**What is your favorite thing to do with your brother:** Either simply hanging around our house when I am home, or bothering our younger sisters; it is a tough call.

**If you met someone who just found out that their brother had Barth syndrome, what would you say to them that might make them feel better:** I would make sure they understood they weren't alone, and that even if you don't know it there are always people thinking about you and your family.

**What does BSF mean to you? Why is it important to you:** While the Foundation is especially great for my brother and the rest of the Barth boys, I also enjoy being able to meet new people, and learn more about Barth syndrome.



# The Barth Syndrome Trust

## Annual Report - 2006 - Third successful year

By Michaela Damin, Chair, Barth Syndrome Trust



### Save the date!

Our 2007 Clinic and Gathering are scheduled for Friday 6 July (clinic) and Saturday 7 July (gathering) in Bristol. Please contact the Trust for more details.

**T**his past year has been an extremely busy and productive year. Our trustees, volunteers and family members have all worked very hard to ensure growth and sustainability of the organisation. As importantly, if not more so, we have continued to make a difference in the lives of boys affected by Barth syndrome. As the mother of a child who has Barth syndrome and as the Chairperson of the group, I would like to extend my deepest thanks to everyone who has contributed to our ongoing success.

### MAIN ACHIEVEMENTS IN 2006

#### Awareness

##### *New medical brochure*

With the help of Dr. Colin Steward and his team, we created a new medical brochure which is being distributed to health care professionals. This brochure has already been warmly received as a source of much needed information for those diagnosing and treating affected children.

##### *Direct Mail campaigns and one-on-one relationships*

A direct mail campaign to targeted doctors has yielded positive results. We have had contact from doctors who are treating children with BTHS and requests to be added to our newsletter mailing list.

##### *Working with Jeans for Genes (JAG) and the Genetic Interest Group (GIG) on a Family Route Map Project*

The Barth Syndrome Trust was one of six charities selected for this project. Together with GIG, we are creating a vital Family Route Map for all children affected by Barth syndrome. The aim of the project is to help patients and families to access services and information that is currently available. Expected completion date: 2007

##### *Working with the Children's Heart Federation*

On 14th February 2006, together with other member groups, we attended a reception at the House of Commons where the theme of the day was Every Heart Child Matters.

We believe very strongly in working with like-minded groups and increasing our impact by pooling resources.

##### *Media Coverage*

This was mainly in the local newspapers in Hampshire. Reports covered fundraising events, Every Heart Child Matters and Jeans for Genes. In every case there was some description of the symptoms of Barth syndrome. One of our boys was featured in the Spring/Summer Newsletter of the Heart Transplant Families Together group.

#### Website

We worked alongside the International Barth Syndrome Foundation to help create a new international website ([www.barthsyndrome.org](http://www.barthsyndrome.org)) in 2006. We also host a regional UK and European website ([www.barthsyndrome.org.uk](http://www.barthsyndrome.org.uk)). Essential information has been translated into several European languages.

#### Family Services

Our Family Services team consists of four dedicated family members who are able to deal compassionately with other affected families. They remain in regular contact with families and they provide empathy, support and information to member families.

#### Gathering

On 7th October 2006, we held our first ever 'gathering', on the day after the Bristol Clinic. The families and some volunteers met in Bristol with Dr. Steward and his family as the guests of honour. The next day all visited *At Bristol*, courtesy of the management there. Meeting Dr. Steward, the families and especially the boys has given the volunteers a better understanding of their needs and a new interest in the work. We are grateful to the Children's Heart Federation and the London Law Trust who financed the weekend through their grant programs.

(Cont'd on page 19)



# The Barth Syndrome Trust ~ Third successful year

(Cont'd from page 19)

## Science and Medicine

### BSF International Conference 2006

We encouraged doctors and scientists in Europe to attend our International Conference in July 2006 in Florida, USA. Eight specialists from The Netherlands, the UK, France and Germany took part in the clinics, and the scientific and medical sessions in Florida.

## Looking Ahead - Future Plans

### What do we hope to achieve in the next year?

- We are planning to roll out our International Medical Database and Biobank and allow UK families to contribute data to encourage better understanding and future research into Barth syndrome.
- We will be holding our Annual Clinic and Gathering in Bristol on 6 -7th July 2007.
- We will be organising a European Gathering in Belgium on 2 September 2007.
- We will step up our efforts to provide essential information to doctors and to find more families.
- We will participate in the European Eurordis project (a 2-year project to determine the best way to care for families affected by rare disorders)
- We will finalise the Family Route Map project with the Genetic Interest Group (GIG) and Jeans for Genes (J4G).
- We will finalise comprehensive policies and procedures for all volunteers.
- We will actively seek partners (through grant making organisations and individual funders and fundraisers) in order to achieve our stated objectives.

### European Family Gathering

Sunday 2nd September 2007  
~11am onwards

Open to all European families  
Bokrijk Outdoor Folk Museum & Park,  
Belgium

Entrance fees and lunch to be provided  
by Barth Syndrome Trust.

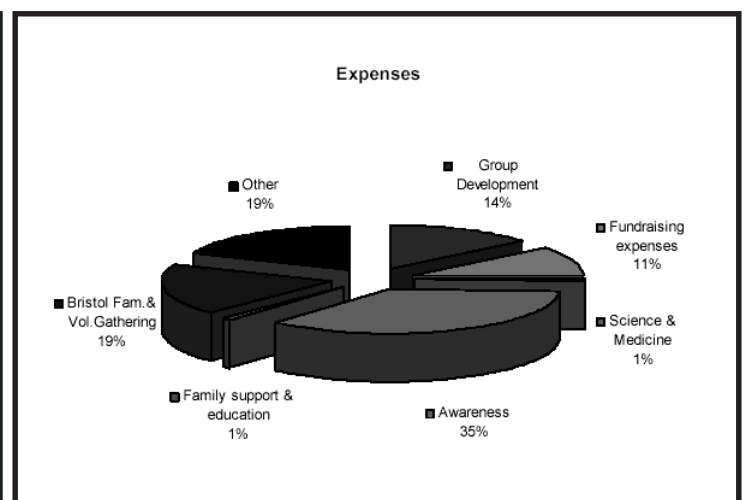
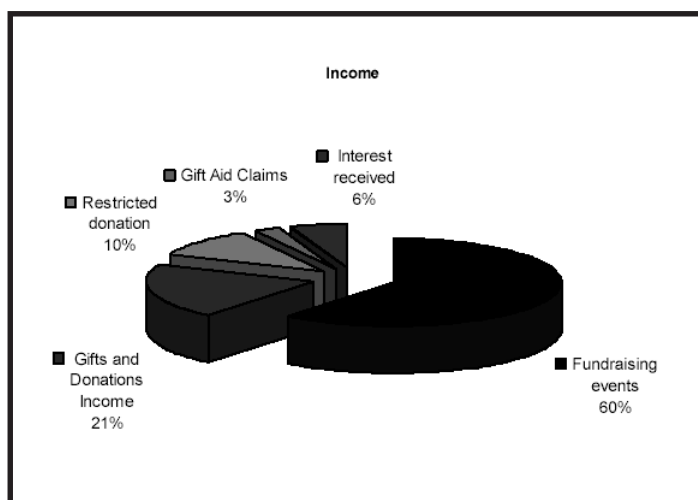
Please contact us for more details.

## Financial Review

### Receipts and Payments Accounts

See Independent Examiner's Report on the accounts by Keens Shay Keens, Milton Keynes.

Year	31 Dec 2006	31 Dec 2005
Receipts	£29,044	£ 23,039
Assets	£ -	£ 1,100
Payments	£ 8,892	£ 5,202
Net	£20,152	£ 17,837



# Barth Syndrome Foundation of Canada

## Canada has banner year!

*By Lynn Elwood, President, Barth Syndrome Foundation of Canada*

Looking back on the close of 2006, it was another exciting year for Barth Syndrome Foundation of Canada. As our financials show, we continued to make gains in fundraising and closed the year with a healthy bank account which will enable us to do much more.

At our recent Annual General Meeting in April, two of our board members (Lynn Elwood and Chris Hope) were re-elected as President and Treasurer for a further two-year term. It is a privilege to work with such a dedicated board and executive as we continue to grow our organization.

Year ending December 31, 2006		
Opening Balance		\$37,872
Revenue	\$50,485	
Expenses	\$22,247	
Excess Revenue over Expense		\$28,238
Closing Balance		<u>\$66,110</u>



**BACK (L-R):** Bill Clelland, Maureen Pitkethly, Audrey Hintze, Cathy Ritter, Chris Hope, Les Morris, Wayne Bridger  
**FRONT (L-R):** Janet Warren, Lois Galbraith, Marjorie Sawh, Janet Humphries

In 2006 we were not able to realize all our program goals, not because we lacked the funds but because we did not have enough of our most precious asset: people. Our group of volunteers has been working hard and made many amazing things happen, but we discovered that we could have done more if we had a broader group of volunteers. We have begun to address this and have held our first volunteer workshop, hosted by Janet Warren. This warm and comfortable afternoon resulted in the expansion of our volunteer group, and these energetic recruits have already been a tremendous help. At left is a picture from our first volunteer workshop. We are planning further meetings with the next one in the fall of 2007.

We have embarked on a major goal, funding of scientific research benefiting Barth syndrome. In 2007 we are funding \$20,000 US of the grant entitled: Cholesterol Metabolism in Barth Syndrome, which was awarded to Grant Hatch, PhD. This is an exciting beginning for us, and we hope to participate in BSF's 2008 grant cycle.



**(L-R)** Dr. Matt Toth, Jason Downard, Shelley Bowen, Lynn Elwood, and Cathy Ritter at the Pediatric Academic Societies' conference in Toronto, Canada.

During 2006 we had several ads and calendar events posted in *The Medical Post*, a newspaper seen by many physicians in Canada. Very late in 2006 we received the offer we've been working on for so long – the chance to have a feature article in this publication. Lois, several members of BSF's Scientific Medical Advisory Board and our members worked over Christmas to help with background and quotes, and the result was a full page colour article with reference on the front page. This is a big step in awareness amongst physicians in Canada and we're very grateful to everyone who helped. We have the rights to reprint this article so if you would like a copy please contact us.

We have also participated in another significant awareness initiative in 2007. We funded and jointly staffed a booth at the Pediatric Academic Societies' conference in Toronto from May 5-7. It was a pleasure to see Shelley and Matt join this conference, and many of the members of the executive (and Friends) helped too. Thank you especially to Robert Hope and Jason Downard for helping in the booth. Several of the doctors commented on how helpful it was to meet someone with Barth syndrome.

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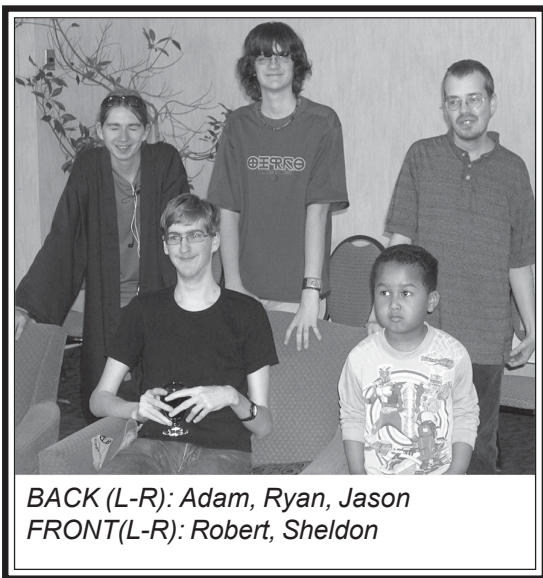
# BSF of Canada has banner year!

(Cont'd from page 21)

In February BSF Canada was a key organizer and partial sponsor of a multi-disciplinary clinic at the Hospital for Sick Children in Toronto. This clinic focused on the hematological aspects of Barth syndrome, and several other conditions. The meeting was led by Dr. Dror from the Hospital for Sick Children. Four of our families attended (approximately 10 people), and there were 70-80 people in total at the clinic. One of our affected men, Robert Hope, gave an outstanding talk on Barth syndrome. Goals of the day included examining some of the common issues and treatments across the conditions, transitions in care from pediatric to adult care, and issues of daily living. We found common bonds with the other groups and have made some good contacts. The families especially appreciated the opportunity to ask our young men questions about the issues they face daily.



*In February BSF Canada was a key organizer of and partial sponsor of a multi-disciplinary clinic at the Hospital for Sick Children in Toronto. This clinic focused on the hematological aspects of Barth syndrome, and several other conditions.*



*BACK (L-R): Adam, Ryan, Jason  
FRONT(L-R): Robert, Sheldon*

We try to combine our Annual General Meeting (AGM) with a family gathering. It is hard to get everyone from the diverse geography together, but with Toronto being somewhat central we are able to meet with some of the families in person. This April's AGM saw us getting together with several of the families including the Hones from Saskatchewan. After our meeting and recreation time for the young men, we shared a dinner and laughs together. We did not manage a full group picture, but here are some of the Barth individuals who attended the meeting. Thanks to all who made the trip to join us. It is always so wonderful to see everyone in person.

Our fundraising activities in 2007 are off to a good start. The letter campaign has already received over 50 responses. Our *Change for Barth* campaign has expanded beyond the month of May for those that want to keep saving change for the whole year. We have also benefited from a private fundraiser by Marj Sawh. On September 10 we will have another golf tournament at Tangle Creek in Barrie, ON. Visitors from all regions are welcome to join. We are also planning an end-of-year fundraiser. With all that we are doing in our programs, these fundraising events are more important than ever.

Given the pace of projects so far in 2007, it is a good thing we are joined by a growing group of volunteers. We're looking forward to the road ahead as we work through our 2-year program goals. Please feel free to get in touch with any of us if you would like to find out more about our program activities or plans. There are teams on each of these programs, and here are the current program leaders for Canada:

Cathy Ritter, Family Services  
Lynn Elwood, Science & Medicine  
Chris Hope, Awareness  
Lynn Elwood, Charity Support  
Cathy Ritter, Fundraising  
Lois Galbraith, Volunteers

[critter@barthsyndrome.ca](mailto:critter@barthsyndrome.ca)  
[lilwood@barthsyndrome.ca](mailto:lilwood@barthsyndrome.ca)  
[chope@barthsyndrome.ca](mailto:chope@barthsyndrome.ca)  
[lilwood@barthsyndrome.ca](mailto:lilwood@barthsyndrome.ca)  
[critter@barthsyndrome.ca](mailto:critter@barthsyndrome.ca)  
[lgalbraith@barthsyndrome.ca](mailto:lgalbraith@barthsyndrome.ca)

We look forward to sharing more news with you in the fall newsletter, but please feel free to contact us at any time.



# Barth Trust of South Africa

## Highlights of 2006 accomplishments

By Jeannette Thorpe, Chair, Barth Trust South Africa



*Jeannette Thorpe, Chair, Barth Trust South Africa, with her son, Ben.*

It's just amazing how quickly time goes by – we are almost half way through 2007 and the pace does not seem to be slowing down. Reflection and consolidation give one time to take a deep breath and see life in perspective once again, showing that the year 2006 marked some significant growth for the Barth Trust of South Africa:

### **Science and Medicine**

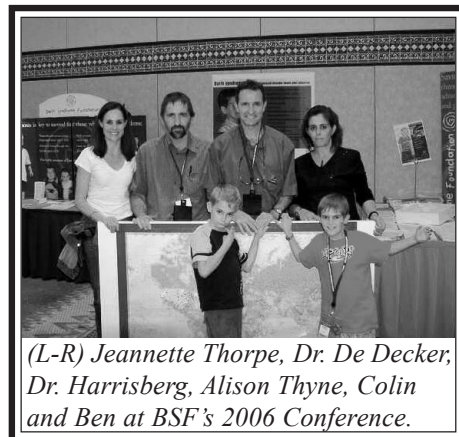
In July 2006, Dr. De Decker and Dr. Harrisberg were the first South African doctors to attend BSF's Scientific and Medical Conference in the USA. They were given an insight into Barth syndrome on various levels: in a practical way, through attending and observing the clinics and interacting with other families and affected boys and men; and clinically, through the many highly informative presentations.

The genetic testing facility was moved from Durban to Cape Town. Our long term vision is to develop a Centre of Excellence for Barth syndrome in South Africa and it therefore made sense to consolidate our resources.

### **Awareness**

Doctors De Decker and Harrisberg gave the first clinical presentation on Barth syndrome in South Africa at the South African Heart Congress on October 31, 2006 in Somerset West, Cape Town. Presentations at congresses are a particularly effective means of raising awareness.

Public awareness was also raised by a detailed newspaper article published in three Provinces in South Africa, as well as by a radio interview in which Dr. De Decker participated at the end of 2006.



*(L-R) Jeannette Thorpe, Dr. De Decker, Dr. Harrisberg, Alison Thyne, Colin and Ben at BSF's 2006 Conference.*

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*“Dr. Jeff Harrisburg and I were privileged to attend the 3<sup>rd</sup> Barth Syndrome Foundation Conference in Florida, USA. It proved to offer an enthralling window into a complex and enigmatic disorder, which we are only just beginning to understand. The quality and depth of the research to uncover the syndrome’s secrets are astounding. More importantly, we came to know a group of parents who through enthusiasm, resilience, and hard work, have managed to energize the process of understanding Barth syndrome. They have succeeded in changing the perception of the disorder from a singular rarity to one which must be considered in every boy who has cardiomyopathy. We should be grateful to assist with raising awareness of Barth syndrome in South Africa to this level of recognition!”*

*~ Dr. Rik De Decker, Red Cross Children’s Hospital, Cape Town*

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Save the Date!

# Barth Syndrome Foundation International Scientific/Medical and Family Conference

(Including Poster Sessions & Clinics)  
**July 21-26, 2008**

*Bellevue Biltmore Resort ~ Clearwater, Florida*

"I think the scientific and medical sessions of the last conference were the most mature ones BSF has organized. Not only did they include a large number of researchers with very diverse backgrounds, they also initiated a new way of interaction between these scientists. ..." ~ *Michael Schlame, MD, New York*

"The 2006 Barth Syndrome Conference was a life-changing experience for our entire family. We now feel that we are a part of something so much bigger and more powerful than Barth syndrome itself! We are so thankful for this group and what it has already meant in the life of our son. We are counting the days until the 2008 Conference." ~ *Floyd Family, Georgia*

"A joint meeting of families, scientists, researchers, clinicians and educators. All groups gain from this encounter: the families in getting comfort from one another and hope from research progress heard first-hand; the doctors and scientists in witnessing the plight of patients and families." ~ *Salvatore DiMauro, MD, New York*

"BSF's 2006 Conference completely exceeded our expectations. We were struck by the dedication and commitment of the families and doctors." ~ *Strain Family, Australia*

"As a physician interested in the genetic basis of pediatric heart diseases, such as cardiomyopathy, my attendance at BSF's conference was invaluable in learning about patients with this genetic disorder and about scientific progress into the mechanisms of disease and genotype-phenotype correlations." ~ *Arnold Strauss, MD, Ohio*

"We now know how important it is for everyone to try to attend the conferences. They are extremely inspirational." ~ *Galley Family, Australia*

Please bookmark BSF's website [www.barthsyndrome.org](http://www.barthsyndrome.org) for up-to-date information.

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



Barth Syndrome  
Foundation

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

Phone: (850) 223-1128  
Facsimile: (850) 223-3911  
E-mail: [bsfinfo@barthsyndrome.org](mailto:bsfinfo@barthsyndrome.org)  
Website: [www.barthsyndrome.org](http://www.barthsyndrome.org)