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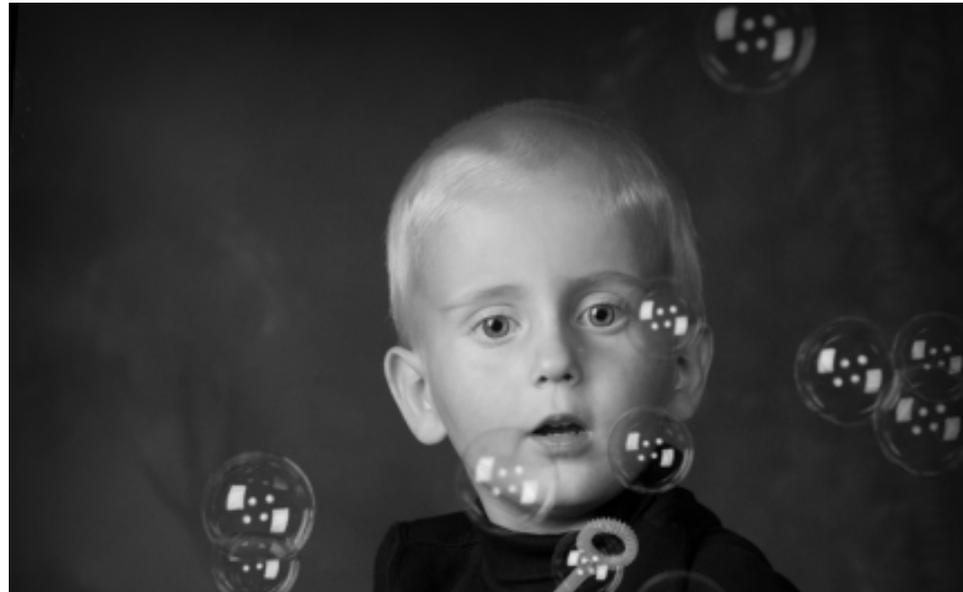


Photo by Amanda Clark

Research into Barth syndrome BSF initiates Barth Syndrome Registry and DNA Bank!

By Carolyn T. Spencer, M.D., Assistant Professor, Director, Pediatric Echocardiography Laboratory, University of Florida College of Medicine, Gainesville, Florida

The 2006 Barth Syndrome Foundation Conference was a tremendous success for those involved in research on Barth syndrome. Not only were researchers able to work with families and boys with Barth syndrome to collect important information in

many areas (cardiology, neurology, psychology, and exercise/physical therapy), but this conference marked the initiation of the Barth Syndrome Registry and DNA Bank. For both clinical and basic science, researchers interested in Barth syndrome, and for the Foundation itself, this is an important milestone.

(Cont'd on page 5)

Families bonding for the cause Conference brings together eleven first-time attendees

By Shelia Mann, Chair, Family Services

The Barth Syndrome Foundation International Scientific, Medical and Family Conferences are truly one of the greatest tools to deliver a better understanding of Barth syndrome to families, physicians, scientists and researchers. Each topic provides affected families with crucial information about this complex disorder.

The conferences also offer affected families one additional, very important component. It brings families from around the world together to bond and form friendships that will have an everlasting impact on their lives. The sharing of knowledge and wisdom from past experiences can shed new light on issues about the disorder that might never have been looked at before. So thank you to

(Cont'd on page 8)

**Scientific and Medical
Advisory Board**

Richard I. Kelley, MD, PhD –

Chairman
Division of Metabolism
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Peter G. Barth, MD, PhD

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Children's Hospital
Boston, Massachusetts
Clinical Research, Genzyme Corp.
Cambridge, Massachusetts

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Neurology
Columbia University College of
Physicians and Surgeons
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Wilmington, Delaware

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Biological Sciences
Wayne State University
Detroit, Michigan

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Lipid Lipoprotein Research
University of Manitoba
Winnipeg, Canada

Michael Schlame, MD

Anesthesiology
NYU School of Medicine
New York, New York

**Colin G. Steward, MA, BM, BCh,
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
Houston, Texas

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.
New York

What is passion?

By Valerie ("Shelley") Bowen, President

Dear Friends,

Every six months it is my privilege to write a letter to you, the BSF community. Even though I know this is coming, I struggle to find the right words to convey my gratitude to so many people I have never met. Those who know me well realize just how much this matters to me.

This is an exciting season in the evolution of BSF: we have closed an important chapter in our history. We have, in fact, surpassed all of the objectives outlined in our first five-year strategic plan. Our vision of the future, "*A world in which no one will suffer or perish from Barth syndrome,*" is built on painstaking planning, evaluation of best practices, seeking out opportunities, dedication, and teamwork. However, at the core of all of this, is passion.

So, what is passion? When you really dig into the etymology of the word, the synonyms, "zeal, ardor, enthusiasm and fervor," seem inadequate. Over time "passion" has been used too often, diminishing its intensity. Passion is not fleeting, and all of us who make up BSF are driven by it.

On Saturday, November 4, 2006, I witnessed passion in person when I attended the Ironman event in Florida for the first time. When I arrived, Gary Rodbell greeted me at the 78-mile marker, seated on his bicycle looking strong and determined. He had already completed the 2.4 mile swim, had another 34 miles to ride, and then an entire 26.2-mile marathon to run. His wife, Collette and daughter Julia were there to cheer him throughout. Along with them were family members and others, including his long-time friend Steve McCurdy. Together with his friends John Steigerwald and Matt Karp, Gary had created Team Will to participate in this event to raise funds for BSF. Training for these events was very grueling, and required sacrifices of time and energy that I am sure were difficult at times.

This was Gary's third Ironman for BSF, and at the age of 53, after being hit by a car during his training, this would be the last of these events for him. Gary's passion for BSF started with the love for one boy, Will McCurdy, but his passion grew as he began to learn about all of the others who have Barth syndrome. My son is one of those who benefited from Gary's passion, and I was thrilled to be there to support him during this arduous event.

At mile marker 127.5 the smile on Gary's face faded; he was clearly hurting. But his passion would not allow him to slow down. In the end Gary completed the race with a finish that was his personal best. And, after months of raising funds for the triathlon "Team Will" claimed first place in the Janus Charity Challenge. As I sat on the sidelines I watched many contestants overcome enormous obstacles: one blind athlete was tethered to a friend because he needed a guide throughout the race; another challenger who had been diagnosed with terminal cancer seven years ago responded by training for the Ironman. Each contestant brought with them a passion that fueled them through this agonizing contest. I watched as Matt Karp ran to the finish line with a smile on his face, and the next day I dropped him off at the airport so that he could return to his family.

The Ironman made me consider the many people involved in BSF, some of whom I have never met. I thought of the many donors listed at the end of this newsletter who believe in our cause. They are believers because someone who was passionate about our organization asked them to help. I thought of all of the hours devoted to BSF by dedicated volunteers. These contributions of all kinds move us ever closer to our vision of a world where Barth syndrome no longer represents a threat.

(Cont'd on page 3)

(Cont'd from page 2)

“I am glad that I was able to be a part of something so meaningful to so many people. In doing all of our parts ...

a community of support joined hands with incredible strength to try and make the world a better place.”

**Matt Karp
2006 Ironman**

As the president of this great organization, I would be the very first to say that all of our success is the product of extraordinary teamwork. My passion for this organization is not unique. It is the dedication of all of our staff, volunteers, donors and friends that has made us able to meet the ambitious goals we identified five years ago.

As I write to you, we are beginning to identify the goals and workplans for another five years. We believe that all the work we have done up to now, with your help, is increasing our understanding of how to help our boys and their families. I would like to thank every person who has helped us on this journey, and I would like to say again that I believe that the world will be different as a result of our endeavors.

Respectfully,



BSF surpasses its first five-year strategic goals

BSF's Leadership team met earlier this month in Prosperity, SC to celebrate the successful completion of its first 5-year plan and to map out the next 5 years!



BSF Science Director hired!

By Katherine R. McCurdy, VP, Science and Medicine

In the last newsletter, I wrote that BSF was conducting a nation-wide search for a highly qualified and dedicated Ph.D. to serve as the Foundation's professional Science Director. We were astonished and heartened by how many wonderful candidates applied for this position (we received nearly 50 good applications for our one position!). It made our job of selection more difficult in some ways, but it also was fabulous to have such extraordinary choices. In this issue, I am thrilled to announce that we have hired Matthew J. Toth, Ph.D. in this role, and he is just the right person to join our team.

(Cont'd on page 4)

For all correspondence, including information, please contact:

Board of Directors

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

Stephen Kugelmann, VP, Awareness
skugelmann@barthsyndrome.org

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

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

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

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

Additional Contacts

Matthew J. Toth, Ph.D., 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
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 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.



BSF warmly welcomes Matthew J. Toth, Ph.D. as Science Director.

(Cont'd from page 3)

Matt completed his Ph.D. in microbiology at the Massachusetts Institute of Technology (MIT) in 1988. Since then, he has worked in the pharmaceutical industry on many projects and products that are relevant to Barth syndrome. He has

BSF Science Director hired

worked on lipoproteins and with metabolic conditions, and he has done projects with *Drosophila* and knock-out mice. At Novartis Pharmaceuticals Corp. (formerly Ciba-Geigy Corp.), Matt was a laboratory head for Metabolic and Cardiovascular Disease projects. Most recently, at Amicus Therapeutics, he was the Director of Pre-Clinical Research and led the company's work on developing products for several rare diseases (Pompe Disease and Gaucher Disease). The list could go on, but as is clear to see, Matt brings a wealth of knowledge and experience to BSF that will be invaluable to our progress.

Matt also has the right personal attributes to fit in

beautifully at BSF. He is a devoted family man; he and his wife, Marilyn, live in New Jersey with their two teenage children, a daughter and a son. Furthermore, he believes in fostering excitement in science among young people and cares deeply about contributing back to his community. As a volunteer, Matt has been a lead organizer of the North Jersey Regional Science Fair for many years now.

Matt's interest in cutting edge research with a focus on development of products that can help patients, as well as his love of science education and his strong desire to make a difference in the lives of others make him an ideal addition to the BSF team. He

joined BSF in time to attend the International Science, Medical and Family Barth conference in July and was able not only to learn a great deal about the disorder, but also to meet many of the researchers, clinicians and families associated with our Foundation.

After the conference, Matt said, ***"I hope very much that I can live up to the expectations of the boys. I would like nothing more than to make a real difference to their quality of life and to their future."*** He already has shown commitment to our cause and compassion for our boys and young men. We feel very fortunate that he has joined us.



Board of Directors announces new addition!

By Stephen B. McCurdy, Chairman, BSF Board of Directors

The Board of BSF is very pleased to announce that Susan S. Osnos has accepted our invitation to join the Board of the Barth Syndrome Foundation with a 3-year term that runs until April of 2009.

Susan Osnos is already known to our volunteer leaders and is no stranger to BSF. In fact, she attended one of our first meetings (as a public relations advisor) when a small group gathered to decide how to establish a foundation in 2000. Susan was instrumental to our success in getting the "Saving Michael Bowen" story placed in Readers Digest, and equally critical to the recent 30-minute story on *Mystery Diagnosis* that continues to run on the Discovery Health Channel in

the United States. Susan attended the Volunteer Enrichment sessions in South Carolina in 2005 and 2006 to help train our leaders in the "art" of public relations for BSF. She also joined us in Orlando for our most recent BSF Conference, and has been a faithful contributor to BSF since its very first year.

Susan's qualifications are well suited to BSF. She helped to grow a highly successful not-for-profit – Human Rights Watch – a global organization that has become a strong, respected, and highly influential advocate for human rights around the world. She joined Human Rights Watch in 1984 soon after its creation, and helped to shape its public image for 13 years as Communications

Director, followed by three years as its Associate Executive Director. She serves on the Board of Words Without Borders, is a member of the Council on Foreign Relations, and continues to serve on the Advisory Committee, Women's Rights Division of Human Rights Watch.

Susan is also a cousin of Will McCurdy, and has watched one of our Barth families learn to cope with this disorder for the last twenty years. She blends an understanding of, and sensitivity to, the concerns of the Barth families, with decades of experience in growing and running an international non-profit organization. Susan is uniquely qualified to help us increase awareness, expand our



organization, and begin to find and blend non-family resources with our growing family base of volunteers.

Susan lives in Greenwich, Connecticut with her husband Peter, Founder of Public Affairs, a book publishing company. They have two adult children – Katherine Sanford, mom of Ben and Peter, and Evan Osnos – Beijing Bureau Chief for the Chicago Tribune.

Research into Barth syndrome

BSF Initiates the Barth Syndrome Registry and DNA Bank

(Cont'd from Cover page)

Just as in the 2002 and 2004 BSF conferences, clinical data were collected during the 2006 Conference to continue to follow these boys over time. The results of the first analysis of Barth syndrome patients from the 2002 and 2004 conferences were recently published (Spencer, et al., *Cardiac and Clinical Phenotype in Barth Syndrome. Pediatrics.* 2006 Aug;118(2):e337-46). The initial group consisted of 34 patients. Since that time, we have expanded the total group to 47 patients. Of this group, 11 have had consistent 6 year follow-up and 23 additional patients have had at least 2 year follow-up. Thirty-three boys with Barth syndrome were evaluated at BSF's 2006 Conference, including 12 new boys.

Tests performed at the 2006 Conference included 29 echocardiograms, 29 ECGs, 12 metabolic exercise tests, 11 exercise echocardiograms, 12 microvolt tests, 27 sets of laboratory tests, and 23 subjects with muscle strength testing. Additionally, 7 healthy boys (mostly brothers of those with Barth syndrome) participated in a separate study as a control group, including metabolic exercise testing and muscle strength testing. The researchers and staff participating in this effort were from the University of Florida Department of Pediatrics, Children's Hospital Department of Cardiology, and Washington University School of Medicine.

The 2006 BSF Conference also served as the launch of the Barth Syndrome Registry and DNA Bank. Although the Registry and DNA bank will be housed at the University of Florida, it will be available to researchers everywhere. The goal is to develop a complete database of medical information of boys with Barth syndrome, and this information will be linked to the DNA/blood sample to allow researchers to better understand the

clinical and genetic abnormalities in Barth syndrome. All medical information and blood samples will be labeled by a special code that does not identify the enrolled patient by name.

Currently there are 35 boys with Barth syndrome enrolled in the Registry (although we are still waiting on some Registry Intake Questionnaires from enrolled families!), with 23 samples in the DNA bank, and 21 sets of lymphoblast lines. An advisory committee has been set up to oversee the Registry and DNA Bank (see below). We hope to have the Registry and DNA Bank operational for outside investigators in early 2007. For more information, please contact: barthregistry@ufl.edu, or call Dr. Spencer at 352-392-6431.



Drs. Rik De Decker (Cape Town, South Africa), Carolyn Spencer (University of Florida, Gainesville, FL), and Jeffrey Harrisberg (Gauteng, South Africa) collaborate during BSF's 2006 Conference.



Barth Syndrome Registry and DNA Bank **Advisory Committee**

Members: Principal Investigator, BSF Scientific and Medical Advisory Board Chair, BSF Board of Directors appointee, and other members appointed by the Principal Investigator

Principal Investigator: Carolyn Spencer, MD, University of Florida

Members:

Richard I. Kelley, MD, PhD, Johns Hopkins University
Chair, BSF Scientific Medical Advisory Board

Gerald F. Cox, MD, PhD, Children's Hospital Boston and Genzyme Corp.

Michael Schlame, MD, New York University School of Medicine

Colin G. Steward, MA, BM, BCh, FRCP, FRCPCH, PhD, Bristol Royal
Hospital for Children

Shelley Bowen, President, Barth Syndrome Foundation, Inc.

BSF 2006 Conference

Delivers Scientific advances and excitement

By Matthew J. Toth, PhD, Science Director



An international, dedicated group of scientists, researchers, physicians, clinicians and educators gather at BSF's 2006 Scientific, Medical and Family Conference to advance the knowledge of all aspects of Barth syndrome.

In July 2006, a remarkable scientific, medical and family conference on Barth syndrome was held at Lake Buena Vista, Florida. About 35 scientists and clinicians with approximately 149 family members joined together from all over the world to learn about this disease and to hear what advances, both clinically and scientifically, are being made to overcome it. Characteristically, each scientific session was led off with a short presentation by a parent of a Barth boy, and culminated with the final session being introduced by a 20-year old Barth individual.

The Conference was supported by a generous R-13 grant from the National Heart Lung and Blood Institute (NIH) and the Office of Rare Diseases (NIH), which recognized the meeting as important for the advancement of knowledge about all aspects of this disorder. There is not space available to list all of the terrific speakers and topics that were presented among this collaboration of colleagues, but the highlights are described briefly within this article.

Gene Products and Biochemistry

The first session, which focused on the "Gene Products and Biochemistry of Barth Syndrome", was chaired by Miriam Greenberg, PhD (Wayne State) and Michael Schlame, MD (NYU). Mutations in the *tafazzin* gene are the cause of Barth syndrome, and the first session covered cellular and animal models where the *tafazzin* gene is deleted or deficient in its expression. Mingdong Ren, PhD (NYU) and Michael Schlame, MD discussed the fruit fly mutant of *tafazzin* which has reduced flying and climbing activities. These traits resonate with the fatigue that many Barth boys suffer. Dr. Greenberg showed that adding certain metals to the growth media of yeast mutants deficient in the *tafazzin* pathway can restore the mutant to a normal state. Arnold Strauss, MD (Vanderbilt) discussed the difficulties in making a *tafazzin* knockout line of mice. With animal and cellular models, we should be able to probe what other genes are altered when *tafazzin* itself is defective. This may lead to better ways of treating symptoms, or maybe to finding a mechanism-based way to overcome the dysfunction caused by *tafazzin* mutations.

New Frontiers of BTHS Science

The second session, which focused on "New Frontiers of Barth Syndrome Science", was chaired by Richard Kelley, MD, PhD (Johns Hopkins). In this session two researchers, Francois Bernier, MD, FRCPC, FCCMG (U of Calgary) and Robert Jensen, PhD (Johns Hopkins), discussed the Barth syndrome-like mitochondrial disease found in the Hutterite community of Canada. These patients suffer dilated cardiomyopathy and other maladies similar to Barth patients. The gene responsible for the problem encodes a protein needed for proper protein movement into the mitochondria. Understanding how a protein import problem resembles a *tafazzin* mutation should lead us to a better understanding of both conditions. Grant Hatch, PhD (U of Manitoba) spoke about cardiolipin and phospholipid remodeling using fibroblasts from Barth syndrome patients. The changes observed in these lipids are uniquely characteristic of Barth syndrome.

Cardiac Issues

The third session, which focused on "Cardiac Issues of Barth Syndrome", was chaired by Jeffrey Towbin, MD (Texas Children's), who discussed the clinical aspects of left ventricular noncompaction in Barth patients originally described with dilated cardiomyopathy and hypertrophic cardiomyopathy. Dr. Towbin also described the "undulating" course of many Barth patients who dramatically improve their cardiac condition to the point of being taken off the heart transplant list and of discontinuing cardiac medication. Carolyn Spencer, MD and Randall Bryant, MD (U of Florida) presented clinical data they have gathered from a large cohort of Barth patients over the last several years, with support of a BSF grant. Arnold Strauss, MD (Vanderbilt) described how the knockdown of *tafazzin* expression in zebra fish embryos using morpholino oligonucleotides caused severe cardiac abnormalities which are consistent with the cardiac problems found in Barth patients. (Cont'd on page 7)

(Cont'd from page 6)

Hematological Aspects

The fourth session, which focused on the "Hematological Aspects of Barth Syndrome", was chaired by Colin Steward, MA, BM, BCh, FRCP, FRCPCH, PhD (Bristol Royal), who discussed the cycling nature (usually 19-21 days) of neutropenia found in many Barth patients, and the difficulty of clinically recognizing this condition. Dr. Steward showed that very low doses of granulocyte colony stimulating factor, given at the proper time in the cycle, are effective in mitigating some of the effects of the neutropenia without risking serious side effects. David Dale, MD (U Washington) then added some information that he has garnered about Barth patients from the Severe Chronic Neutropenia International Registry that he oversees. Taco Kuijpers, MD, PhD (Emma Children's) showed cardiolipin abnormalities in the neutrophils of Barth patients and a large amount of Annexin-5 binding. Normally Annexin-5 binding signals cell death or apoptosis, but these Barth neutrophils were not recognized by the macrophages that would normally destroy them. Willem Kulik, PhD (U of Amsterdam) and colleagues presented a poster describing a microassay technique for determining cardiolipin levels from only a bloodspot sample, which when validated, should provide an easy way to monitor this very distinctive hallmark of Barth syndrome.

Barth Syndrome Registry and DNA Bank

In the last scientific session, Carolyn Spencer, MD (U of Florida) introduced the Medical Database and Biorepository for Barth patient clinical samples and medical records, housed at the University of Florida. This resource will be a significant way to leverage the brainpower of the entire scientific and medical community.

The Chairs of the sessions concluded the Conference by discussing with the audience several areas that need advancement. The neutropenia suffered by the Barth boys needs to be understood. How cardiolipin is utilized within the mitochondria of the cell, and why some Barth boys have more severe symptoms than others are also perplexing issues. How does the *tafazzin* gene product contribute to the cardiolipin abnormalities? These unanswered questions go to the core of the mechanism that is dysfunctional in Barth syndrome and are a preview of what meeting attendees will be looking forward to hearing about at the next conference.

Post-conference

Post-conference surveys of the participants rated this meeting as very positive in many aspects such as collaborations with colleagues, gaining new information, and generating new ideas to take back with them. Many of the presenters had publications about their work appear in print shortly after this conference. In addition, the number of publications concerned with Barth syndrome has increased every year for the past several years, demonstrating an increasing awareness in the scientific/medical community.

Summary

In summary, this Third International Conference was a great success, distinguished by the scientific advances that were presented and by the collaborative atmosphere where new relationships were begun and old ones were reinvigorated. The conference also showed that more needs to be done among the researchers, clinicians, and families to get us to the place where we all want to be.



(L-R) Dr. Colin G. Steward, Chair of the panel focusing on the hematological aspects of BTSH, along with presenters Drs. David C. Dale and Taco Kuijpers, discuss the cycling nature of neutropenia in Barth syndrome, and the difficulty of clinically recognizing this condition.

"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.

The speakers were asked to answer specific questions related to Barth syndrome. As each presenter did so from his or her own angle, new ideas emerged, which will have an impact on research in the coming years. In that sense, the conference has accelerated our understanding of the disorder.

All of us who are studying Barth syndrome look forward to the BSF-sponsored conferences, as they provide to us a crucial platform to exchange information.

Perhaps one of the most significant accomplishments of the conferences is that the number of research groups that are working on Barth syndrome has increased every year."
~ Michael Schlame, MD



Families bonding for the cause

BSF Conference brings together eleven first-time attendees!

(Cont'd from Cover)

those who were present and for sharing your knowledge and experiences with BSF.

The 2006 Third International Scientific, Medical and Family Conference was a huge success on many levels due to the commitment and organization from the 2006 Conference Committee. The committee of volunteers consisted of Jan Kugelmann, Shelley Bowen, Kate McCurdy, Lynda Sedefian, Chris Hope, Shelia Mann, Alanna Layton, Lynn Elwood, Michaela Damin, Jeannette Thorpe, Joyce Lochner, and Lois Galbraith. BSF would like to thank each of them and the many other volunteers involved in organizing the conference. BSF would also like to thank Dr. Richard Kelley, BSF's Scientific Medical Advisory Board, and the many dedicated physicians and researchers who committed their time and knowledge to our cause.

It was truly an international event, and BSF was delighted to see families represented from Scotland, England, South Africa, Australia, Canada, and the United States. There were a total of 35 Barth families represented and a total of 34 affected Barth individuals present. We also had a record-breaking turn out of 11 first-time attendees. Those families were able to bond and meet other Barth individuals and families face-to-face for the very first time. The relationships that were formed between families made attending the Conference a life-changing event.

Barth Clinics

The 2006 Conference was remarkable, emotional, interactive, and thought provoking. The knowledge gained during the multi-disciplinary clinics will help further facilitate

research and future treatment strategies. The assessments and clinical data gained from each affected Barth individual will support the Barth Syndrome Registry and DNA Bank, which is critical to future research. During these clinics a total of 34 affected Barth individuals were assessed in the following specialties: cardiology, neurology, genetics, education, psychosocial, OT/PT, and nutrition. Families had the unique opportunity to consult with world experts in all components of the disorder. They were able to address specific concerns that related to their affected Barth individual. What more could you ask for?

Family / Medical/Scientific Sessions

The conference provided two tracks of meetings for its attendees. One track focused on families of affected individuals, while a simultaneous track focused on the scientific and medical aspects of Barth syndrome. In an effort to give our affected families the tools to become the most effective advocates for their Barth individual, the Family Sessions included expert presentations and panel discussions which focused on providing the most up-to-date information on current research advances, the

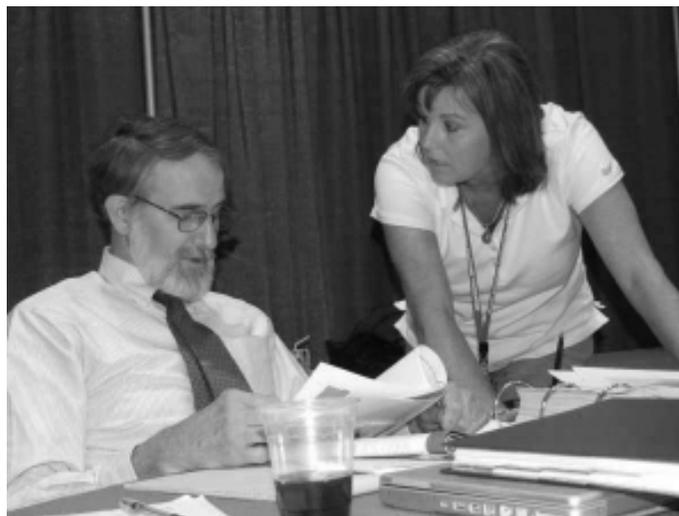
clinical aspects of Barth syndrome (BTHS), day-to-day aspects of BTHS, and current resource information about BTHS. The panel format gave families the opportunity to ask questions, enabling them to gain a deeper understanding about this complex disorder. On the last day of the conference, breakout sessions were utilized which focused on age-specific clinical and psychosocial aspects, as well as education and transitional aspects.

Youth Sessions

BSF also offered sessions for the affected Barth youth and their siblings (ages 8 and up). These very special meetings that only occur every two years provide a wonderful time for these young men, boys and siblings to bond and form lasting relationships. They truly have a feeling of belonging and a feeling that they are not alone in this journey. The memories created and love that is shared give them new hope for a bright future!

Memories that will last a lifetime

The social and bonding opportunities for attendees were endless throughout the conference. The planned Friday Night Social provided an evening of socialization among families, physicians, scientists and researchers. Entertainment, music and an international theme set the stage for a fun-filled, emotional evening. Dancing, laughing and sharing the emotions of this relaxed evening with new formed friends and partners for life created memories that will last a lifetime. Attending a BSF conference is truly an everlasting experience for the entire Barth family. The spirit in the room was filled with hope for a healthier future for present and future Barth generations.



Shelia Mann consults with Dr. Richard Kelley during the Family Track of BSF's 2006 Conference.



From birth to independence

The role of therapy

By Karin Watson, MPT; Lynn Hancock, OTR/L; Jeanette Van Duyne, PT; Bobbie King, OTR



Occupational therapists Lynn Hancock and Bobbie King work with Ben, age 12, during the OT/PT clinics held at BSF's 2006 Conference.

“These assessments allowed us to determine areas of weakness compared to age-matched peers, as well as determine trends or patterns within the developmental progress of boys with Barth syndrome.”

any given sample. This raises the question of whether the boys are “true” left-handers or whether it is an adapted response to poor muscle strength and lack of endurance.

From our findings we strongly emphasize the importance of physical and occupational therapy.

Early Years – Birth to Four
During the “*Early Years*,” therapy should focus on achieving motor milestones, developing strength in the shoulders, trunk and hips, and creating habits for an active and healthy lifestyle.

As Physical and Occupational Therapists that have had the opportunity to work with only one amazing boy with Barth syndrome, we greatly enjoyed participating in the 2006 International Barth Conference and meeting all the other amazing boys.

Our purpose during the Conference was two-fold. First, we wanted to assess the children using functional tests (standing from the floor, push-ups, sit-ups, step-ups, writing) and using a standardized test – the *Bruiniks-Osteretsky Motor Proficiency*. This test was administered to assess motor functioning of children ages 4 ½ to 14 ½. These assessments allowed us to determine areas of weakness compared to age-matched peers, as well as determine trends or patterns within the developmental progress of boys with Barth syndrome. Second, we wanted to provide a variety of home programs and strategies to assist each family.

So what did we find and what do we suggest?

We discovered that we couldn't get the little boys to slow down, and we had a hard time getting the big ones to wind up!

All of the boys demonstrated very good balance and eye-hand coordination. We determined that those with Barth syndrome have decreased upper body strength, which results in sitting intolerance and poor posture (forward shoulders and head), thus affecting their ability to attend to school-related tasks. They also presented with a decrease in trunk and hip strength, which impacts their ability to run for speed, jump for height or distance, and to stand up from the floor without the use of hands.

Additionally, many of the boys have adapted pencil grasps due to decreased hand strength, and were often observed to switch hands frequently during writing and fine motor activities. Handwriting fatigue was an issue for at least 95% of the boys. Approximately 50% of the boys were left-handed. This is an unusually high number for such a small sample. The norm would be 10-12% of

Honeymoon Phase – Five to Eleven
When entering the “*Honeymoon Phase*,” emphasis should be placed on success in the classroom. This includes proper seating, functional grasp when writing, and introduction of keyboarding. The boys should continue activities that promote overall strength and endurance.

Barth Resurfaces – Twelve to Fifteen
As “*Barth Resurfaces*,” importance should be placed on keeping the boys moving by motivating them to participate in motor activities and sports to their ability, such as martial arts. During this time keyboarding skills become more important to succeed in the classroom.

Independence - Sixteen and Older
The “*Independence Phase*” places focus on self-directed activities that encourage strengthening, posture, endurance, and lifelong fitness.

We thank you again for allowing us to be part of the Barth community and hope to see you at the next conference.



BSF fosters a teamwork approach

Working toward empowering our future

By Jon Rosenshine, M.A., M.Ed.

In the Youth Sessions at the Barth Syndrome Conference this past summer, we tried to respond to the feedback we received from the boys two years ago, while trying to make the sessions as informative and fun for the boys as possible. We had our home base in the Coronado T conference room where we had games, snacks, and chairs, as well as copious beanbags and blankets for comfortable lounging. Time to “kick back, relax, and chill out” was built into the schedule.



The Barth boys/young men participate in teambuilding exercises during BSF's 2006 Conference.



Our Barth boys and young men participating in a “Speed Connecting” exercise during BSF's 2006 Conference.



Meet a few of our Barth youth (ages 12-15), together with Jon Rosenshine, during the Youth Sessions of BSF's 2006 Conference.

The boys had a vigorous and honest discussion with Dr. Eric Storch about issues of bullying, and they received continued education from Drs. Miriam Greenberg and Iris Gonzalez on Barth research and the genetics of Barth syndrome. They also enjoyed demonstrations of various martial arts after which Bill Knauer led them in a lesson of the first Tai Chi form.

Team building and group cohesion among the boys was an important goal of the week, and to that end we conducted Speed Connecting exercises in which the boys sat in rows facing each other and engaged each other in short one-on-one discussions based on specific questions supplied to them. Dr. Roberto Canessa's keynote address was geared especially towards group dynamics in crisis situations, and the older boys had the opportunity to learn from Dr. Canessa about his extraordinary story of his and his teammates' survival after being stranded in the Andes Mountains for 72 days.

Finally, the experience was tied together and recorded by the boys and their cameras. Each boy had the opportunity to capture images of his experiences, of the Coronado Springs Resort, and of his friends. These images were uploaded and organized into a slideshow that the boys presented to the assembled conference on the last day. This year's conference continued the success we have had in the past by giving the boys a safe and fun environment to be together, to share their experiences, and to build their support network among themselves.



Barth children and young men representing Scotland, South Africa, Australia, Canada, and the United States, met at BSF's 2006 Conference to connect with old friends and establish new ones along the way.



Our future leaders (age 16+) who attended BSF's 2006 Conference.



BSF's 2006 Conference also involved specific activities for our siblings of Barth individuals. Old and new friendships were nurtured.



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Recently added peer-reviewed articles

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DVD'S on Sale Now!

Thanks for your interest in the Barth Syndrome Foundation 2006 Conference DVDs. Each session - Family or Scientific & Medical - contains nearly 12 hours of indexed and chaptered presentations from leading physicians, scientists, researchers, therapists and parents. You will quickly be brought up to speed on the research and information known about Barth syndrome, as well as how other families are managing their child's health. You can purchase the Family Sessions, Scientific & Medical Sessions, or both. Each set comes with a BONUS DVD that includes interviews with some of the boys and young men and the Discovery Channel segment from *Mystery Diagnosis*.

"2006 is our second conference with BSF and I just want to express my thanks and gratitude to everyone for welcoming us and allowing us to take part in more than just the work at hand. This is truly a rewarding job and it's our privilege to help make a difference." ~ Steve Kownacki

To order your DVDs, please visit www.barthsyndrome.org.

Barth timeline:

1981 and 1983

Peter G. Barth, pediatric neurologist in The Netherlands, et al., first fully described an X-linked mitochondrial disease affecting cardiac muscle, skeletal muscle and neutrophil leucocytes.

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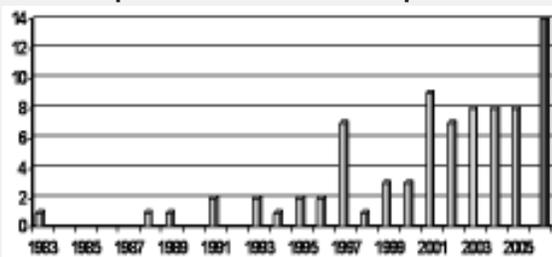
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BSF is making a measurable difference in scientific articles published!

Number of peer-reviewed articles published on Barth syndrome



2006 = partial year actual through Nov. 23, 2006

- Note that the Barth gene was discovered in 1996
- BSF was incorporated in late 2000
- 15 of the 30 papers published since January 2004 have resulted directly from BSF research grant awards

Highlights of Barth syndrome research

2000
Peter Vreken et al. demonstrated that tafazzin is involved in cardiolipin remodeling in Barth fibroblasts.
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2004
Arnold W. Strauss et al., and Mauro Degli Esposti et al., independently created zebrafish knock-in models of Barth syndrome; Strauss demonstrated that G4.5 gene is essential for normal cardiac development in zebrafish.

2004 (cont'd)
Iris L. Gonzalez presented data in 2004 that was then published in 2005 that only two functional forms of G4.5 mRNA exist (delta 5 and full-length) in humans; also noted that exon 5 does not exist in yeast or rodents and that the full ability to splice developed only after evolutionary split from Old World monkeys, but is important to humans.
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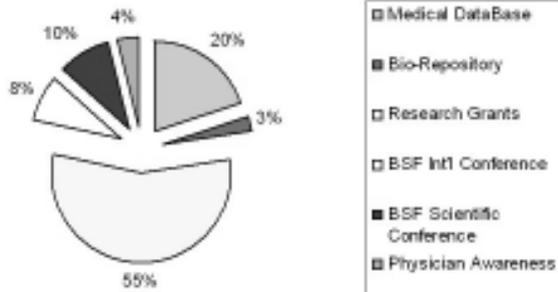
2006 (cont'd)
Michael Schlame et al. created a Drosophila model of Barth syndrome.
Xu Y, Condell M, Plesken H, Edelman-Novemsky I, Ma J, Ren M, Schlame M. **A Drosophila Model of Barth Syndrome.** Proc Natl Acad Sci USA. 2006 Jul 19; [Epub ahead of print].

Carolyn T. Spencer et al. documented the cardiac and clinical phenotype in Barth syndrome.
Spencer CT, Bryant RM, Day J, Gonzalez IL, Colan SD, Thompson WR, Berthy J, Redfearn SP, Byrne BJ. **Cardiac and Clinical Phenotype in Barth Syndrome.** Pediatrics. 2006 Jul 17; [Epub ahead of print].

Claypool et al. showed that yeast tafazzin protein localizes to both membranes that face the intramembrane space in the mitochondria, and BTHS-causing mutations modeled in yeast display mislocalization of the tafazzin protein.
Claypool SM, McCaffery JM, Koehler CM. **Mitochondrial mislocalization and altered assembly of a cluster of Barth syndrome mutant tafazzins.** J Cell Biol. 2006 Jul 31;174(3):379-90.

In 2006, BSF established the Barth Syndrome Foundation Science and Medicine Fund

This \$6 Million fund will support research, create opportunities for collaboration, and gather and organize the data and tissue samples needed by the researchers for the next ten years. \$2.0M has been raised to date...



Barth trek: The next generation

By Donna Strain, Parent of Barth Child, Australia



The Strain Family, from Australia, attended BSF's 2006 conference for the first time!

I started to read about Barth syndrome; cardiomyopathy, neutropenia, and low muscle tone; it frightened me so much I turned off the computer. When I eventually felt a little stronger, I filled out the online form and asked to join. Even though I was made very welcome, I was not ready for the listserv. It was too much too soon. I was grieving so much I couldn't face or take in all of this information, I felt physically ill. Again I turned the computer off, and I could not go back to it for quite a while. It wasn't that I didn't want to know. I did. I just wasn't ready to cope with all of these posts, detailing all of the frightening things that my son was more than likely going to face.

Barth syndrome has caused immeasurable grief in my family. When I first fell pregnant I knew that there was a long history of boys dying very young which was possibly due to a heart condition, but we had no diagnosis. Cameron was my first child and he was born in January 1999. By March that year he was in heart failure and we were devastated. To make matters worse the geneticist told us he suspected Barth syndrome, and with our family history we shouldn't expect Cameron to live out of infancy. We felt numb, dazed and confused; none of the doctors could tell us much about Barth syndrome, so I turned to the internet and found the Barth Syndrome Foundation.

When I finally returned to the listserv I found it extremely useful. Even though I was too shy to contribute, I would read the posts every day and use the information. The 2006 Conference was approaching and we really felt Cameron needed to meet some other boys with Barth syndrome, as he was beginning to become very frustrated with himself and his self-esteem was plummeting.

We took the plunge and went to the Conference. Cameron was very excited and I was very nervous, but we are so glad we did it. From the moment Cameron met another boy with Barth syndrome he lit up; it was amazing to watch. He had the time of his life, and

"BSF's 2006 Conference completely exceeded our expectations. We were struck by the dedication and commitment of the families and doctors."

he has made some wonderful friends whom he talks about every day. My husband, Fraser, loved the Conference. He learnt so much about Barth syndrome and loved meeting the boys, doctors and the families.

A homecoming

For me, it was a homecoming. I finally met people who understood what I had been through, and I realized that the feelings of guilt, and sometimes unbearable sadness were normal, and it was such a relief. Over that period of six days I think I experienced every emotion possible; grief, hope, love, relief, fear, to name a few, but I did it with people who understood. The Conference completely exceeded our expectations. We were struck by the dedication and commitment of the families and doctors. We met some wonderful people at the conference, people who will always be dear to us and who we hope to see again at the next one in 2008!



Farewell to my hero...

By Cameron, age 7, Barth Child, Australia

Steve Irwin ("Crocodile Hunter") was my Hero. I am sad that he had to die. He was a great wild life icon, and he looked after wildlife a lot. He had a passion for wild life, and whenever there was an animal in trouble he was there. I loved visiting Steve at the Australia Zoo, and the last time I went I got to meet him. He was really nice and he was just like he was on the documentaries. I will always remember him, and when I grow up I want to be just like him. I love you Steve, farewell. ~ **Cameron**

Cameron (right), waits patiently for his turn to ride the croc while visiting the Australia Zoo!



Revisiting BSF's Conference

An awesome and truly inspirational experience

By Annie Galley, Parent of Affected Individual, Australia



Warm hellos to everyone. My name is Annie Galley. I have been asked to write on our experiences of revisiting BSF's Scientific/Medical & Family Conference, which is hosted every two years, and the impact that these conferences have had on our lives.

Firstly I will tell you a little about our family and how we became a member of this extremely important group of warm, caring people.

We live on the Gold Coast in Australia. There is myself, hubby Rod, our son Matt, 16, who has Barth syndrome, and our two daughters Courtney, 9, and Chloe, 8. Matt is from my first marriage, and was diagnosed when he was 18 months. My brother, Garry Wakefield, who is 28, also has Barth syndrome, and has had a heart transplant. My mother, Jean Wakefield, and I are carriers of the Barth gene. My sister, however, is not a carrier.

Our son Matt has been up and down like most of our boys, and is currently classed as 'stable', but is going through a few heart problems that we are trying to deal with. We are looking at possibly going onto the transplant list in

December/January, if Matt continues to decline. He is currently on Lanoxin, Captopril, Carvedilol, and aspirin, along with daily injections of GCSF.

Now, in our experience as a 'Barth Syndrome Family', having the Barth Syndrome Foundation in our lives has been a "Godsend."

Attending the first Barth Conference

Matt, my mum, Garry and I were fortunate enough, thanks to our local communities, to attend the 1st Barth Syndrome Conference back in 2000, which was held in Baltimore, MD, USA. It was a dream come true, and we were amazed at how kind, loving, and friendly everyone was. The meetings were great; we were able to consult with expert doctors who were able to answer our questions. It was only a small group at that time, but we all knew there had to be other families that didn't know how to find our group, and this is where and how the foundation started, with Shelley Bowen, Sue Wilkins, and Anna Dunn, taking the leading roles, determined to find other families and grow to where we are now.

Revisiting BSF's Family Conference

The 2006 Barth Syndrome Conference, in one word, was "**AWESOME.**" We didn't think we could make it, but we knew how important it was for Matt and us as a family, so we turned once again to our local community, and thanks to their help and support we were able to attend.

As I mentioned, the Conference was "AWESOME." Having attended BSF's first-ever Conference in 2000, and then having the opportunity to attend the 2006 Conference, I was completely overwhelmed and extremely impressed with how much they have changed, and excited by how far the Foundation has come. There were so many families all just as keen and eager to be there to

hear and learn everything there was to know about Barth syndrome. We all came together to help our sons.

The clinics are so important for us and the doctors, scientists, and researchers, as gathering and testing as many boys as possible will surely help with research and, hopefully, one day a CURE.

Medically, attending the Conference is extremely important, but on a personal level we found the Conference just as important. To be around other families that know exactly what you are going through and feeling is priceless. It really helps just to talk and have someone there that knows what you are talking about. Plus, seeing the other boys of all ages is so amazing! They are all such special boys and young men. Rod and I gained a lot from the Conference, and I was so glad we made the effort to attend. Before we knew we could make it, Matt didn't really want to go, as he is extremely shy. But once the Conference was over, he actually thanked me so much for getting him there, as he absolutely loved it. He made some great friends, and he said it was so good to sit and talk with the other boys about Barth syndrome and how it affects him, and they knew exactly what he meant. He didn't want the Conference to end, which is huge for Matt.

One other thing that hugely impressed me was the Family Social Night. I couldn't get over how the doctors, scientists, and researchers got involved, dancing and mingling with the families. It was such a great thing to see. Where do you ever see doctors and families interacting and having fun? We are truly one big 'family' that is striving to make our sons' lives the best we can.

We now know how important it is for everyone to try to attend the conferences. They are extremely inspirational!! We will be trying our best to attend every two years.



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.



Benjamin, age 2, Texas



Colin, age 9, S. Africa



Jared, age 12, Canada



Darryl, age 24, Pennsylvania

'Faces' of Barth syndrome

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.

Barth syndrome listserv

An amazing tool that glues us together

By Jay Randell, Father of Two Barth Boys

I remember the day as clear as a bell. I was driving in my car and my cellular phone rings. On the caller ID I see the name of my geneticist calling. Two of our boys have just been through numerous hospital stays in the PICU at two hospitals, diagnoses of congestive heart failure with ejection fractions of 6%, cardiac catheterizations, etc.. We all know the story! At the time we had no answers and we were convinced that two of our sons were dying and no one knew why. As was routine, I answered the phone with bated breath but without hesitation. "We have a diagnosis! Your sons' tests came back positive for Barth syndrome." Your sons are number 84 and 85 of the global diagnosed cases, and the organization will contact you shortly. That was it... he had no more information for me. I felt lost and alone in my car. Eighty-five cases diagnosed globally, and no one had any answers. I was told to wait for some "lady" whose son had this disease to call me. (Sorry for the bluntness

Shelley. The compliment is coming). I could not see at the time just how much these few people really did know, and who was this lady and what was she going to tell me that my doctor could not!!!



Now how precious are these two brothers!! Sitting together during a portion of the the clinics that were held at BSF's 2006 conference, they were determined that they were going to see Disney World, and were getting ready to drive themselves to see Mickey!

Now almost two years later I realize that all that I know about Barth syndrome has come from the Barth Syndrome Foundation's listserv and the organization. We have received critical information via the listserv, ranging from corn starch dosing, to captopril concentration regulations, being cautious about discontinuing their digoxin, and that the way my boys "drink" salt is not bad for them. I could not have learned any of this from any of my physicians. The listserv has become our lifeline. Just knowing that this tool is there for my questions and concerns is a comfort. It has become my first resource before we go to our doctor, or even to use it to advise our doctors how to proceed.

What would any of us do without that "lady" who called us the first time and welcomed us into the family?

The listserv is our families' lifeline and glue that helps us grow and care for our boys together.



Firefighters of the Reedy Creek Fire Department were determined to make Jeremiah's wish come true... to see a real fire engine up close and personal!

***"I came for a job,
and I left having
my heart touched
by a very
special young man!"***



BSF Ironmen bring home the gold!

By Stephen B. McCurdy, Chief Financial Officer



Gary stops for a little encouragement from his wife, Collette, at the 2006 Florida Ironman.

BSF's Ironmen achieve personal best !

BSF's Ironman, Gary Rodbell, and his friend Matt Karp, had a good day on Sunday, November 4th. A very long day, but a very good day! Starting at 7AM and in the midst of 2,300 other aspiring Ironmen, Gary and Matt ran into the Gulf of Mexico to begin the 2.4 mile swim, the Ironman's first event. Some 90 minutes later each emerged to begin the 112 mile bike section of the race, followed by a full 26 mile marathon. Thirteen hours and 21 minutes after he started, having never run a full marathon let alone an Ironman before, Matt Karp finished the race. Despite a bruised knee and a 53-year-old body, Gary finished his last Ironman race with a personal best time of 15 hours and 51 minutes. Both men were exuberant and exhausted.

Gary Rodbell wins 2006 Janus Charity Challenge

But the crowning moment of the competition may actually have been achieved on Friday when it became clear that Gary had won the Janus Charity Challenge by raising more than \$284,000 from almost 300 donors for the Barth Syndrome Foundation – the most raised for a charity by any athlete in the Panama City Ironman.

Gary had come in second twice before in the Janus Challenge by raising \$75,000 for BSF with John Steigerwald in Ironman Florida in 2002, and \$150,000 with John Steigerwald and Tim Monetti in Ironman Wisconsin in 2004. Gary set the bar high in 2006, at a seemingly impossible \$300,000. And then, like a true Ironman, he went for it and never lost faith or gave up. As a result of Gary's "win", Janus Funds will contribute an additional \$10,000 to BSF.

This year, Gary has started another tradition. As a tune up, he recruited eight other athletes to race in the Westchester, NY Triathlon (a sort of mini-Ironman) in September and to raise money for BSF and the Janus Charity Challenge. In addition to Matt Karp, Gary was joined by Dr. Bob Buly, Paul Epstein, Rich Gerszberg, Mike Hennessey, Joanne Jensen, Angelo Mancino, Amy Rosen and John Steigerwald,. These amazing athletes swam, biked and ran individually or in relays under the name "Team Will", after Will McCurdy, a young man with Barth syndrome from Larchmont, NY and a close friend of Gary. Gary and Team Will are committed to return to the Westchester Triathlon next year with a larger group to compete again and continue to raise much-needed funds for BSF.



Gary and Team Will are committed to return in 2007 to raise funds for BSF!



Gary and Matt, BSF's Ironmen, take a moment to reflect at breakfast the morning following the Ironman.

After returning from the Ironman and after reading the many e-mails that poured into his mail box from Barth families around the world, Gary wrote the following:

"Crossing the Ironman finish line is like no other feeling in the world. Knowing that I take with me on that one day all the Boys as well is something that I find difficult to articulate. There are so many ups and downs during the race. I find myself wanting to just stop and call it a day after being out there so long. I felt this way coming back to the transition at the end of the bicycling and as the deadline was approaching. The sky was getting dark and off in the distance the "Full Moon" was coming up. It was truly a sight to behold. I thought about the Boys and I thought about the finish line. I knew right then and there whatever it took I would do it.

I just wanted to thank all of you at the Barth Syndrome Foundation for giving me the opportunity once again to bring the Boys along for the day. I and the rest of our Ironmen do not know or understand the pain and suffering so many of the families feel day to day. For me it is a privilege and an honor to run this race for Barth. November 4th was a day I will take with me for the rest of my life."

~ Gary Rodbell



(L-R) Timmy (17), Aldo (14) and Jack (13), a few of our Barth boys, joined in and bowled for Barth.



Joyce Lochner, Kate McCurdy, and Lynda Sedefian display the Barth quilt and matching pillow that was raffled at BSF's Social Night.

Bowling for Barth

On Saturday, October 14, 2006, Liz and John Higgins hosted The Third Annual Barth Syndrome Bowling fundraiser at the Frontier Lanes Bowling Center in Warwick, NY. Among the attendees were two "Barth" families: The Dunn family (Aldo) and the Monahan family (Timmy). Both funds and awareness were raised, and great fun was had by all. Mark your calendars for next year's event, which will be held on Saturday, October 13, 2007.



Steve McCurdy, along with friends and parishioners of St. John's Episcopal Church (Larchmont, NY) raise funds to support BSF.

BSF Conference receives financial support from the US National Institutes of Science

BSF's R-13 scientific conference grant application to the NIH received one of the highest scores possible, and won \$35,000 of combined support from the Office of Rare Diseases and the National Heart, Lung and Blood Institute. The institutes recognized the quality of the application, the science to be explored at the Conference, and the credibility of the Barth Syndrome Foundation as conference sponsor. Credit goes to Sue Wilkins and her daughter, Jess Wiederspan, with the able assistance of Dr. Richard Kelley (Chairman of BSF's Scientific and Medical Advisory Board) and Jackie Pittman at Johns Hopkins in Baltimore, Kate McCurdy and the rest of the Conference Organizing Committee – Dr. Miriam Greenberg, Dr. Michael Schlame, Dr. Colin Steward and Dr. Jeffrey Towbin.

Easter Appeal raises \$53,755 for BSF Science and Medicine Fund

Eighty six generous parishioners of St. John's Episcopal Church in Larchmont, New York made contributions to BSF during Pentecost, which corresponds to the "Birth of Barth" month in May. Led by Carla Berry, the Senior Warden of St. John's Vestry, and the Reverend Tom Nicoll, parishioners responded to a letter beautifully written by Dacey Hall, posters featuring portraits of Barth boys, and a talk by Steve McCurdy, a Barth dad and a long time member of St. John's parish. Linked by the McCurdy family, the local St. John's community and the global Barth community found common cause, mutual respect and a deep and abiding appreciation for each other.

Second Barth quilt raffle

Doctors, parents, scientists, friends and families bought raffle tickets and took their chances to win the Barth Quilt at the 2006 BSF Conference. This beautiful, blue quilt was hand made by Joyce Lochner's friend, Nina Rivers, from Albany, New York. Nina also made the first Barth quilt which was auctioned off at the 2004 BSF Conference. A small army of persuasive young Barth boys and their siblings quickly exchanged raffle tickets for large amounts of folding money from the Conference attendees. Alison Thyne took the Barth Quilt home to South Africa and was thrilled to have a piece of Americana as her memento of her 2006 Conference experience.

For information on fundraising activities around the world, please see articles written by BSF's affiliates on pages 20-24.



Barth Syndrome Trust: 'Progress and plans'

By Michaela Damin, Chair, Barth Syndrome Trust

As Christmas approaches, it seems a good time to look back at what we have accomplished and to the future at what we plan to do next. We look to a future where Barth syndrome no longer exists, and we work in the present to make the lives of those who have Barth syndrome a little easier. Firstly though, I would like to thank our trustees and volunteers for bringing a very high standard of expertise and experience to us.



BST volunteers gather at BST Volunteer Workshop.

One of our main jobs is to ensure that we are good stewards and that we are leading the group correctly, efficiently and honestly. As the Chairperson of this group, I pledge my ongoing dedication to all our members. Together, we will continue to share our unique skills in order to further our goals.

I was very touched to see that every affected family attended our recent Clinic and Gathering Weekend, thanks in great part to funding from the Children's Heart Federation and the London Law Trust. It

was wonderful to see several of our very dedicated volunteers there too, volunteers who are not affected by Barth syndrome, but who have become invaluable partners. It has also been

heartening to witness the commitment and collaboration between doctors in Bristol, the United States, and Amsterdam in making this clinic a success. Thank you all.

***“As the
Chairperson
of this group,
I pledge
my ongoing
dedication to all our
members.
Together, we will
continue to share
our unique skills in
order to further
our goals.”***

~ Michaela Damin

individuals in the UK and Europe who range in age from infants to adults in their thirties and forties. We make up a significant section of the total Barth population world-wide.

We will continue to serve our affected families and we are here when they need us. We will continue to raise awareness so that more accurate diagnoses can be made. We will work alongside others in the common furtherance of our goals. And I am confident that we will succeed.

Best wishes for a healthy and happy 2007.

~ Michaela Damin

Europe

It has been another challenging period for this vast and diverse region. Our main achievement has been the translation of the most important information about Barth

syndrome and BSF/BST into six European languages for the new website. Thank you to all who worked so hard to complete this in such a short time.

The Bruge family barbecue in Belgium in September was one of the BST fundraising highlights this year, raising 3500 EUR! Thank you to the Family and everyone who supported this event.

BST's focus in Europe for 2007 will be awareness, partly to highlight the fact that with advances in care and treatment boys with Barth syndrome have a brighter future.

We have also restructured our Family Services. Johan Fioole, who himself has Barth syndrome, is the new contact for Dutch speaking families. We extend a warm welcome to Johan Fioole. (jfioole@barthsyndrome.org). Annick Manton will be the contact for French speaking families. Annick and Veerle van Langendonck are planning Family Gatherings for these two language groups next year.

(Cont'd on page 21)

Barth Syndrome Trust: 'Progress and plans'

(Cont'd from page 20)



“Jo van Loo has been an inspirational partner and friend, and has been instrumental in building the European group.”

~ Michaela Damin

It is with the greatest sadness that we say goodbye to Joke van Loo. She was involved with BSF long before I joined, and she was a founder member of BST in Europe. She has been an inspirational partner and friend, and has been instrumental in building the European group. Jo has resigned as a trustee, but will still be involved to a lesser degree in Europe. We all wish her the very best in her new career.

~ Michaela Damin

Bristol Clinic and Social Day

We travelled from Scotland to attend the Barth Syndrome Clinic and we are so glad that we did! The uplifting feeling that our whole family gets from attending these days makes the journey so worthwhile. We are very grateful to Dr. Colin Steward, Dr. Bev Tsai-Goodman and Michaela Damin for again giving us this unique opportunity. It is rare to meet doctors who know so much about Barth syndrome, so we were not going to miss this chance.

Clinic days can often be stressful for the boys, but I think our son, Alfie, had so much fun in the playroom that he didn't mind the echo, blood tests and ECG. Non-affected family members also had bloods taken to help with research using a cardiopilin comparison test.

The families had group sessions with an endocrinologist, cardiologist, geneticist and gastroenterologist where doctors and families shared information. We appreciated doctors asking families for their experiences and opinions; they showed a genuine interest in Barth syndrome. We came away with more information that we, as parents, can relay back to Alfie's consultants at home.

As always, it is so nice to meet the other affected families, to share laughter, experiences and information. One of the best things about clinic days is that the boys and their siblings can see each other again.

The family and volunteer social day was a lovely idea. It was so nice to meet and thank some of the people who help BST families. My family is deeply grateful to each and every one of the doctors, consultants, volunteers, and all who help with BST. It makes us hopeful of a brighter future. *~ Allanna Anderson, Mother of Alfie (6) and Jay (7)*

Fundraising

Some of our wonderful parents, their families and friends ran half-marathons recently raising awareness and over £2,000. Many thanks to Eleanor, Lorraine, Tommy, David and Dave.

Dave Bull, who completed the Bristol half-marathon despite injuries says, 'It was my way of helping BSF/BST to continue helping new families, and together we hold the key to our future successes.'

Tommy Anderson who ran Glasgow half-marathon wrote, 'The 2006 Conference brought home to me how dedicated BST and BSF are to helping our boys, and this gave me the determination to do well for our extended family'.

Our Open Day at Longstock Water Garden was another event which brought our volunteers and families together to spread awareness and raise funds. There was a wonderful atmosphere amongst helpers and visitors. Our youngest helper, Eleanor (age 12) writes, 'After a few damp days the weather cheered up and far more people visited than we ever expected'.

Thank you to the John Lewis Partnership who own this beautiful garden and all at the Leckford Estate who lovingly care for it.



Families gather at the 2006 Bristol Clinic



Barth Syndrome Foundation of Canada

New frontiers

By Lynn Elwood, President, Barth Syndrome Foundation of Canada



Eager golfers hit the course to raise funds for BSF of Canada at 2nd Annual Golf Tournee.

The six months since the last update have gone by very quickly for most of us on the BSF of Canada executive. As with so many others in the BSF organization, we were all involved in BSF's 2006 International Conference held in July in Orlando, FL. Members of the executive helped in a variety of ways before and during the Conference, and all but one of us were in attendance at the event. The clinics were well attended and the physicians/clinicians were committed, as well as friendly and open. Medical database/biorepository data (samples) were collected, and so another exciting phase begins for Barth syndrome.

Raising awareness

In the area of awareness, we have renewed contact with Children's hospitals across the country and will be sending them an updated package of information. Resource centers and libraries in Children's hospitals are an excellent source for new Barth families, and we will continue to ensure that information about Barth syndrome is available. Each of the hospitals will also receive BSF's newsletters. We are also investigating physicians whose contact information is not current. If you receive our newsletter and expect to move, please send us your new address so we can keep you informed.

Cathy Ritter was asked to speak at the Child Neurology Conference in Pittsburgh, PA this October. This presentation given to international neurology nurses marks the first major initiative to spread awareness among the nursing population. Cathy provided the group with an overview of Barth syndrome, including pathophysiology, incidence, symptoms and treatment; the neurological presentation of the Barth child, and the nursing implications of caring for a child with Barth syndrome. Barth syndrome will also be highlighted on the neurology nursing website for the period of one year.

We are thrilled that we are now ready to fund scientific and medical research from Canada.

Funds have been allocated towards this program, and we are actively seeking an appropriate grant to participate in. More information about the process is available to interested parties (email: info@barthsyndrome.ca), and updates will be sent out when we are successful in finding an appropriate research project. We are extremely proud to be able to undertake this goal so early into our BSF of Canada years.

Our fundraising has grown beyond our expectations again this year. The May 2006 mail campaign was a resounding success. Thank you to all of our friends who responded and helped us to reach \$10,500 in donations. "Change for Barth" was also a successful program, bringing in \$1,700 from an almost completely different set of friends.

In September 2006 we had our second annual golf tournament. The Tangle Creek facility has expanded in the last year, and our tournament grew to match it. This year we had 124 golfers with

us, including three of the Barth young men. All reported having a great day of sport and friendship. We were thrilled with the number of repeat golfers, as well as the many new faces. There were more silent auction items than last year, and there was quite a bidding war over some of the items, including the beautiful quilted wall-hanging donated by Carol Wilks, and the airplane propeller donated by Hope Aero. There was something for everyone – if they were the lucky last bidder. Lois Galbraith, Cathy Ritter, and their team of volunteers did an outstanding job, and we raised \$18,500 to help the Barth Syndrome Foundation of Canada. The raising of awareness of Barth syndrome was equally important.

Planning for the future

During the fall we will participate in the BSF Strategic Planning Volunteer Workshop and share in international plans for the next five years. This will form some of the background for our 2007 program year planning. As we grow, the planning becomes more important and more exciting. We will keep you informed of 2007 plans as they are available.



Jamie Shepherd, Lois Galbraith and Cathy Ritter welcome the 124 golfers to BSF of Canada's 2nd Annual Golf Tournament.

Barth Trust South Africa

Sponsors physicians to attend conference!

By Jeannette Thorpe, Chair, Barth Trust South Africa



(L-R) Jeannette Thorpe, Drs. De Decker and Harrisberg, Alison Thyne, Colin and Ben (front row), all of South Africa, travelled many miles to attend BSF's 2006 conference.

BSF's 2006 Scientific/Medical and Family Conference was, once again, a resounding success. Thank you to all of those involved in putting it together – it was a huge job!! The conference provides a powerful forum for those affected by Barth syndrome and those treating it.

Drs. De Decker and Harrisberg (the first two South African doctors to attend a BSF conference) spent a very productive week observing clinics, learning from the many scientific and medical presentations, speaking with the world's experts on Barth syndrome, and spending time with family members and affected boys/men. They were both very impressed with what they saw and learned and have, in turn, brought this valuable information back to South Africa. They gave their first presentation on Barth syndrome at the end of October at the South African Heart Congress. This was the first medical presentation on this disorder in South Africa. A very exciting step forward! They also plan to submit an article to the South African Medical Journal within the next few months.

After lengthy discussions, we decided to move the genetic testing facility from Durban to Cape Town. Professor Pegoraro (who conducted the first genetic tests for Barth syndrome in South Africa) will continue to support the process. We are grateful to Dr. Tricia Owen and Dr. Peter Berman (of the IMD Laboratory) for agreeing to take over the genetic testing of Barth syndrome. This is an accredited lab. Thank you to Susan Kirwin and her laboratory for providing this lab with all the necessary information.

Now that we have solid structures in place to support physicians who may come across cases of Barth syndrome and families that may be affected by it, we shall again focus on awareness and education for the next couple of months.

I would like to end by thanking Drs. De Decker and Harrisberg again for their tremendous effort of attending BSF's Conference in July (with all the hitches of international travel!) and for their ongoing support here in South Africa!



Thru their very own eyes... *By Ryan, age 15, Barth Individual, Ontario, Canada*



Ryan holds on to special memories of BSF's 2006 conference.

Last July, my mom and I went to the conference in Florida. The conferences are important because they help the research for a cure. But to me, it's all about seeing all the other guys and to get acquainted with all the new people.

To me this is what the conference is about. Not tests or lectures on the syndrome, even though we need to know about it. The best part for me is getting to know people who are affected by the same illness, and sharing all our experiences with them, and showing the younger guys that everything will be fine even though you might not be able to do everything other kids might.

I have special memories of the conference, like when a pile of us went out for dinner and the dance. Most of all I'll remember the time I spent hanging out with everyone. I will also remember the sad time of departing, knowing that we won't be seeing one another for two years. On a personal note, I'd like to say hi to everybody and that I hope you stay well. ~ Ryan Ritter



Sibling spotlight

By Alanna Layton, Sister of Barth Individual

Below are the profiles of three of our fantastic BSF siblings who attended BSF's 2006 International Conference. It is wonderful to see siblings of those affected with Barth syndrome form new friendships among our community of "Barth Families." We strongly believe that these relationships are so meaningful and will be everlasting!



Name: Tamara S.
Age: 14
Address: Queensland, Australia
Affected sibling: Cameron, 7 years

Hobbies: Horse riding, Soccer, surfing

What does the BSF conference mean to you: It is a good chance for my brother to meet other boys with Barth Syndrome and for Donna and Dad to find more ways to help him.

What did you enjoy the most about the conference this year: I found it really rewarding to see Cameron out there meeting and getting to know other boys with BS. I worry about Cameron so it is good to know that they are trying to find a cure. I really enjoyed Disney World it was great fun and it was great to travel to the States and meet people at the conference.

What means the most to you about BSF: I think it is really great that they are trying to find a cure and that there is heaps of support for Cam.

Is there a lesson you learned at the conference that would like to share: I found it really surprising at how many boys are actually affected by BS and at how unknown BS is in the medical field.



Name: Nicky B.
Age: 12
Where do you live: Tampa, Florida
Name and age of affected sibling: Andrew, age 17

Hobbies: Video games, sports

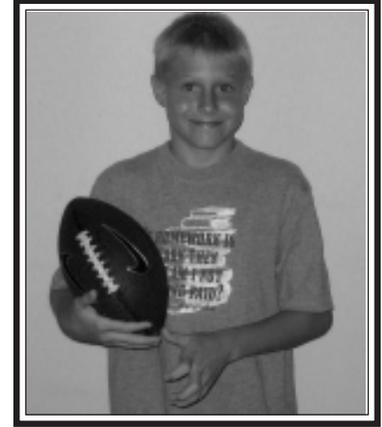
What does BSF conference mean to you?: It means a lot to me because everyone is nice, and helps explain Barth to the families.

What did you enjoy most about the conference this year? The speeches from all the kids.

What means the most to you about BSF?: To know that there are other kids affected by Barth - and knowing that they (the grown-ups) are searching for a cure.

Is there a lesson you learned at the conference that you would like to share?

Never give up, because if you don't give up you'll probably reach your goal.



Name: Benjamin B.
Age: 14
Where do you live: Tampa, Florida
Name and age of affected sibling: Andrew, age 17

Hobbies: Golf, football and all other sports

What does BSF conference mean to you?: The BSF conference means to me a place where people with Barth syndrome and their families come to learn about the disease and how we're trying to come up with a cure.

What did you enjoy most about the conference this year? Seeing all those families coming from all around the world to support their child/brother with Barth syndrome.

What means the most to you about BSF? BSF is so important to me because it shows me that there are people who care about all the kids with this disease and they are striving to find a cure and my brother is one of them and I would love for him to be cured.

Is there a lesson you learned at the conference that you would like to share? The lesson I learned is not to give up and always stick together especially through the hard times.

Power of kindness

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Allman, Peter/Maureen
AME Association of Ontario
American Express Employee Giving
American Express Global Volunteer Action Fund
Amlicke, Thomas/Susan
Anchor Road Stores
Anderson, John & all at British Aerospace, Prestwick
Anderson, Steve / Juhlin, Deborah
Anderson, Tommy Sr. & friends in Porthcawl
Anderson, Tommy/Allanna
Andrews, David/Ellen
Answorth, Dr. Allen
Antomarchi Family
Archer's Eyewear
Architects in Association
Asaff, Ernest/Colette
Aviall Canada Inc.
Azar-Kuhn, Laura
BD Matching Gift Program
Baffa, Ted/Rosemary
Bagnato, Bridget
Bailey, Barbara/Scott
Bancroft, Rev. Frank
Barbier, Etienne
Barrett, James
Bat and Ball Club
Bater, Jennifer N.
Beefy Gang LLC
Behling, Sue
Benevolent and Protective Order of Elks
Berens, Wayne/Harriet
Bergquist, Clay
Berkowitz, Robert
Berridge, Mary
Berry, E.J.
Berry, Rick/Carla
Bez, Ann
Bialo, Katherine/Kenneth
Bienstock, Gregg/Beth
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Bina, David
Bird-Graper, Teddy/Maureen
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Black's Spray Service, Inc.
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Block, David/Sharon
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Blumenfeld, Philip
BMW Canada, Inc.
Boachie-Adjel, Dr. Oheneba
Bob Steele Chevrolet, Inc.
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Bogert, Nicholas/Sally
Bogner, Eric
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Bowen, Shelley/Michael
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Bradley, Robert/Nancy
Bradt, Meg
Brennan, Bridg
Brenner, Andrew/Kathleen
Brevard County Firefighters
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Brooker, Michele/Bradford
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Bucci, Erika/Michael
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Buckeye Florida, L.P.
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Bull, Corey/Kai/Ashley
Bull, Dave/Sarah
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Burdett, Alfred/Norma
Burkett, Marcia/Bradford C.
Burmeister, Charles/Marita
Burns, Geraldine
Burns, Edward
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Bussen-Mayer Engineering
Byrd, Todd/Michelle
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Cahaney, Gail
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Case, Charles I. Jr.
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Cook, Christopher
Cooper, Cindy/Rick
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Cox, Patricia/Gerald
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DeYoung, Edward/Jill
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Dispatch Transportation Corp.
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Kalish, Geoffrey
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Knights of Columbus
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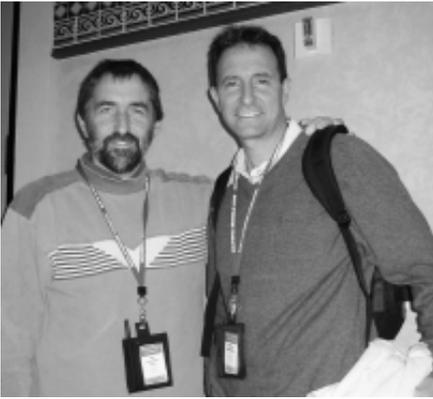
Miller, Gary/Cornelia
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Moncure, Suzanne
Monetti, John/Rejane
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Rader, Stephanie
Randolph, Gregory

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Wilks, Carol/Bruce	Fortier, Jodie	Lyall, Doug	Ryan, Dr. Robert	Kiwanis Club of Provo, Division 10
Williams, Kathleen	Foxon, Kim	Madgett, Roberts, Marlowe, Jackson & Associates	Satula, Deborah	Lochner, Joyce
Williams, Lynn/Robyn	Frost, Tracy	Mah, Dr. Cathryn	Schantzen, Sandy	MacMillan Publishers, Basingstoke
Wilson, Tracy	Gagnon, Lou Ann	Maisenbacher, Melissa	Schlame, Dr. Michael	Microsoft (United Kingdom)
Winner Auto	Galbraith, Lois	Mancino, Angelo	Schroeder, Raghad	Newberry Opera House
Winoker, Sidney/Irma	Ganz, Doug	Mann, Shelia	Schroeder, Wallace	Reedy Creek Emergency Services
Zangara, James/Marie	Gaudin, Herve	Manton, Annick	Scott, Mackey	Rotary Club of Provo, District 5420
	Geigle, Dr. Paula	Manton, Rob	Sedefian, Lynda	Ruzzier, Paolo
	Gemmell, David	Manton, Greg	Segui, Damaris	Shady Grove Radiology
	Gerszberg, Rich	Mantou, Joan	Shaddy, Dr. Robert	Southwest Airlines Civic and Charitable Contributions (Martin, Tracie / Watford, Debbie, Mgrs.)
	Geva, Dr. Tal	Margossian, Renee	Shajer, Esther	Souyri, Francois
	Gilmour, Lisa	Martin, Joy	Shenkman, Dr. Elizabeth	
	Gilmour, Lorraine	Martin, Dr. Rob	Sherbany, Dr. Ariel	
	Gonik, Renato	Martins, Raquel	Sherman, Cathy	
	Gonzalez, Dr. Iris	Maruno, Yuriko	Sheppard, Jamie	
	Gordon, Karen		Sherwood, Dr. Geoff	
			Shirley, Alan/Denise	

Time and Advice
 ASI Production Services, Inc.
 Adelstein, Dr. Robert
 Aikens, Sharonda
 Allen, Dave
 Allison, Mark
 Allison, Terri
 Anderson, Suzie
 Anderson, Tommy/Allanna



(L-R) Drs. De Decker and Harrisberg at BSF's 2006 conference.

Barth syndrome

An enigmatic story

By Dr. Rik De Decker, Red Cross Children's Hospital, Cape Town, S. Africa

I should have guessed from the start that this was to be no ordinary collaboration.

I was attending a cardiology congress in Durban, about 2000km from home in Cape Town. I was idly strolling through the commercial exhibits when a DVD presentation on heart failure caught my eye. The role of carvedilol was being discussed. I blinked, and the sales representative pounced, happy to have another victim. But the DVD was aimed at adult CCF, and, being a pediatric cardiologist, my interest waned. This did not deter the rep. He asked, "As a pediatric cardiologist, do you ever see children with Barth syndrome?" Trying to hide my ignorance, I said, "Not yet, but I'll keep my eyes peeled." I did not add that I would not be able to recognize a Barth syndrome child if he bit me in the kneecap. "OK, but you know that there are only two boys in South Africa with Barth syndrome?" Interesting, but no, I had no idea. I had, I said, to rush off to the next session, and dismissed Barth syndrome as an extreme rarity.

That evening, I visited a close friend, and we, in turn, were invited to dinner at my friend's neighbor. When the neighbor heard that I was in Durban for a heart congress, she immediately said, "Do you ever see children with Barth syndrome?" Almost choking on my dinner, I blurted a confession that I had no idea what she was talking about. "Well, you see", she said, "My best friend, and her sister each have a boy with Barth syndrome." She outlined the salient details of the syndrome, and I was impressed at how well informed she seemed. I gave her the contact details of an "adult" cardiologist in Cape Town who has a strong research interest in cardiomyopathy, suggested that her friend contact him for support and advice, and once again thought no more of it.

Soon after my return to Cape Town, however, I received a call from Jeanette Thorpe, mother of one of the Barth boys. She had contacted the adult cardiologist with the interest in cardiomyopathy, but found that his interest was focused on his chosen field. But, she asked, would I be interested in attending a conference in Florida to learn more about Barth syndrome? She had done her homework, and discovered that I had an interest in cardiogenetics. Did that not include Barth syndrome? Put like that, I realized that she had a point. The mention of cardiogenetics didn't hurt either...

That is how I became involved with the Barth Trust of South Africa, and how Dr. Jeff Harrisberg and I were privileged to attend the 3rd 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, however, 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 to change 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!

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: info@barthsyndrome.org

Website: www.barthsyndrome.org