

## A Life-Changing Impact ... The Journey From England to BSF Conference

By Dave Bull (Bristol, England)



Ashley (13 mths.) in the arms of his father Dave, along with Kai (3 yrs.) and Corey (6 yrs.), in the arms of their mother Sarah

My name is Dave, the father of Corey (6), Kai (3), and Ashley (13 months), and the husband of Sarah. My story starts near enough to the birth of Ashley on September 25, 2004. Ashley had a normal delivery, but even from his first feed he showed no signs of wanting to feed. As the first few weeks went by there seemed to be no real problems, aside from his feeding. By Christmas, we had a real problem with feeding, and Ashley was admitted to the hospital and began nasogastric feeding. After approximately one week, Ashley was diagnosed with

dilated cardiomyopathy (DCM), a heart problem I knew existed because my nephew on my wife's side had it six years previously and needed a heart transplant.

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## The BSF Board Restructures for the Future

By Valerie "Shelley" Bowen, President, and Stephen McCurdy, Chairman, BSF

Since its birth, the Barth Syndrome Foundation has maintained a single minded focus – *Saving lives through education, advances in treatment and pursuit of a cure for Barth syndrome.* As a small foundation with the lives of countless boys at stake, every resource must be put to the best possible use. This basic principle applies to our financial resources, our relationships, our expertise, and our volunteer resources, including our Board members. As the needs of the foundation change, so too must the organization, led by the Board and guided by a set of values that help to keep us on track and reaching the right decisions. This is a story about “finding the right seat on the BSF bus”, and it starts with the Board.

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*Dear Friends,*

*In the past six months The Barth Syndrome Foundation (BSF) has created even more momentum in the growth of our organization and continues to embrace what makes us unique. What makes the core of BSF so unique is our collective resolve to serve all who are affected by this disorder, and our firm belief that we can make a real difference in the lives of all those who are affected by Barth syndrome.*

*We celebrate a number of great strides and accomplishments within BSF and also share tremendous sorrow since our previous newsletter edition. While our accomplishments continue to mark progress within the group, the loss of our children is a painful and important reminder of why we are here. We can never forget that there are children in our midst who are struggling; children who we must help win the battle against this disorder that unites each of us.*

*Our 2004 International BSF Conference, which we held last July, brought a record number of attendees from both the scientific and family community. We simultaneously hosted sessions for the scientific, family, sibling and affected individuals. As I looked around at the time and saw all of these amazing and devoted individuals, I was moved by the progress within our group. For some of us, our first gathering in 2000 was a lifetime ago. In 2000, many of us had never even met another family with this disorder and there was no BSF. In 2004, we were listening to scientists tell us of the progress in research funded by BSF. The US National Institutes of Health sent representatives to attend and speak at our conference.*

*Even though progress has been made within the group, we are reminded that we have no time to waste. We need to do a great deal more for this organization. All of our families are depending on us to work together to make great strides in saving the lives of those who have Barth syndrome. With that thought at the fore, we restructured our board and our operational committees. We took a great leap in embracing the concept of creating a more professional board and cohesively working together to accomplish our goals through strong leadership.*

*We recognize the need for a united stand in our growth. We hosted the most profound Steinhatchee workshop for our volunteer leaders from around the world to date, with a record number of attendees (27 total) including leaders from Canada, the UK and South Africa. Everyone who came to this workshop has made a personal commitment to volunteer to help us attain our goals. I was amazed by the commitment of these men and women who are working toward helping those affected by this disorder.*

*Our BSF community rejoiced in the birth of little Benjamin Holly and continues to share in the profound sorrow with the loss of Gabriel Antomarchi and Benjamin Cooper. The fact that Benjamin Holly and Christian Oldewage could be diagnosed in utero is such a gift. In the celebration of life, we are resolute that every child's life counts. Those who are no longer in our presence will remain in our hearts, and we will continue to forge a legacy on their behalf.*

*Last evening our family celebrated our American Thanksgiving. My husband, Michael and I both acknowledged the influence BSF has had in our lives. We discussed the fact that the very anguish that brought us to our knees has also joined us with comrades of a lifetime. We are all in the trenches together in this battle called Barth syndrome. Won't you join us by taking a more active role? I believe that every one of you will find a place within this organization and that every one who does will be better off for it.*

*Cordially,*



*Shelley Bowen*  
President  
The Barth Syndrome Foundation, Inc.



# BOARD RESTRUCTURES FOR THE FUTURE ...*(Continued from Cover)*

## **Changes to the BSF Board**

Let's start with the changes first and then explain the reasons for them. In a four day retreat for BSF's volunteer leaders held in Steinhatchee, Florida on November 19-22, Shelley Bowen and Steve McCurdy announced several changes to the Board – all designed to prepare the Board and BSF for the future.

- The position of President will become a full-time paid position for BSF, and the Board confirmed that Shelley Bowen will continue in that role
- The Board announced the creation of the new position of Chairman of the Board and elected Steve McCurdy to that position
- Anna Dunn submitted her resignation from the Board and will devote herself to support of the Barth families
- Lynda Sedefian also submitted her resignation from the Board and will serve as a paid executive administrative assistant to BSF and non-voting Secretary to the Board. Lynda will continue as BSF Newsletter Editor
- Steve Kugelmann, Kate McCurdy and Sue Wilkins will continue to serve on the Board, as well as in their current operating roles

## **What is Driving the Need for Change?**

These changes were the result of a continuation of the strategic planning process the Board first undertook in 2001 and has revisited each year since then. As a part of that process in 2004, each Board member was asked to assess his/her strengths and areas of greatest potential contribution to BSF, our mission, vision and goals. Assisted by Gene Early, a professional facilitator who had recently completed a similar task for the Genetic Alliance, the Board met over several days to review and to celebrate the accomplishments of BSF since its creation in late 2000; to consider the tasks still ahead; and to insure that BSF could make the best use of its available resources and successfully attract the additional resources that would be needed in the years to come.

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

All submissions and correspondence regarding the newsletter should be directed to Lynda Sedefian at:  
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\*\* Back issues are also available \*\*

# A Life-Changing Impact ...

## The Journey From England to BSF Conference

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*(Continued from Cover Story)*



*Ashley, 13 months old,  
in the arms of his loving dad (Dave)*

Yes, Sarah and I were devastated, but after a few weeks we were in a "routine" of nasogastric feeding and administering medication. Luckily for us  
B r i s t o l

Children's Hospital is one of the few hospitals that has heard of Barth syndrome. Dr. Colin Steward met with us and described some of the symptoms. Our faces dropped, as deep down we knew he was also describing our second child (Kai). There was so much information to absorb. We were doing our best to learn and understand DCM, then being told about BTHS. We had our family printing information from the Internet, and what we were reading just sounded impossible. We'd have more chance of winning the lottery, so much so that we decided to concentrate on Ashley's heart problem, deep down worrying about Kai.

We are still very new to this, as all of this began in January with testing. In March 2004, Ashley received a positive diagnosis, and it was then recommended that Kai, Corey, Sarah and I be tested. In May of 2004, Kai also received a positive diagnosis of Barth syndrome.

Immediately we were put in contact with Michaela Damin, head of the Barth Syndrome Trust UK, who sent us our new family pack. We were told about the conference and Dr. Steward explained the conference was in seven weeks. He basically said we would learn more in the six days than we would in a lifetime. We wouldn't even travel 20 miles from home for fear of being too far away, let alone 2,000

miles, so we immediately decided not to go. But after thinking about it, we decided we may gain knowledge to help save our children, and realized we could not afford not to go.

We were very fortunate to get a lot of financial help, and we attended the BSF Conference 2004 in Florida. We already knew our lives had changed by having a baby with a heart problem. We knew we had an answer for Kai's delayed motor development and his recurrent sickness as a baby. Back then, our answer was very basic, "Barth syndrome".

Although Ashley was unable to come due to his poor heart, we attended the conference not knowing what to expect. It was the best thing we could have done. Before we attended, we were already in contact with the foundation and had joined the Listserv. We already knew how the foundation had been started and how inviting everyone is, and how, most importantly, the way the boys and the parents bounce off each other, and how many similarities the boys have.

I soon realized that the boys are as individual as any other boy, but there is a special bond that holds us all together, that can, and will, give us answers. Without all the history and enthusiasm of everyone involved with the foundation, our doctors would never have heard of BTHS, and we would probably be none the wiser, until it was too late. We would not have any information about what was wrong with our children, and we would have been left feeling isolated.

I have heard a lot of pre-diagnosed parents say how isolated they felt and no one "gets it". We feel very lucky that we haven't felt this, as there has always been someone there. I know this sounds selfish, but thanks to the parents who have come before us, we have a lot more answers than they

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*"I feel like a small instrument in an orchestra, trying to be heard, with everyone playing, and with the help and support of everyone involved, I believe we will achieve our goal of finding a cure for this condition we call Barth syndrome."*

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did and I feel that we are on a journey, that is going forward, gaining momentum. Without all our information gathered and put together we would know absolutely nothing apart from our own story.

I felt that I learned a lot and gained more understanding from other parents. When we first arrived and met the parents, we felt blessed to meet all these people who we had read about, and had admiration for what they have endured, and had achieved. After getting to know them, I realized that they were ordinary people, parents the same as me. The doctors that were there were very dedicated and professional. They spoke to the parents and boys, and perhaps most importantly they were talking amongst each other.

From a point of perspective from a new parent of Barth syndrome, I feel I have an extended family from all over the world and I feel like a small instrument in an orchestra, trying to be heard, with everyone playing, and with the help and support of everyone involved, I believe we will achieve our goal of finding a cure for this condition we call Barth syndrome.

Nothing can take away the fear we have, not knowing what our boys' future holds, but knowing we are not alone and having the support of the foundation is an overwhelming feeling. I will not forget a comment made to me by a parent who said, "You will not realize just how much you have gained from coming until you've gone home, as it will take weeks for it to settle in." He was right. I can now say I feel that we are on a journey, going forward, gaining momentum. We will definitely be going to the 2006 conference, and would recommend everyone else to go.

I cannot thank everyone involved enough, for your dedication and sheer hard work. Without you, I don't know what our story would have been today!!!



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## HIGHLIGHTS OF 2004 BARTH SYNDROME INTERNATIONAL SCIENTIFIC/ MEDICAL & FAMILY CONFERENCE

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*Reprinted with permission from Pediatric Cardiology Today (August 2004 Issue)*



*Physicians and patients posing at BSF's 2004 International Scientific/Medical & Family Conference*

*By David G. Soergel, M.D., Children's Hospital of Philadelphia*

**B**arth Syndrome (BTHS; MIM #302060) is an uncommon, but probably underdiagnosed, X-linked genetic disorder characterized by dilated cardiomyopathy, skeletal myopathy, failure to thrive and neutropenia. The genetic defect in these patients involves mutation of the TAZ gene (also called G4.5) located at Xq28, which encodes the recently-described tafazzin protein.

The Barth Syndrome International Scientific/Medical and Family Conference, organized by the Barth Syndrome Foundation and held recently in Orlando, FL, brought together the world's experts in the many facets of Barth syndrome (BTHS), including Peter G. Barth, M.D, Ph.D., who first described the disorder. In order to "give a face" to

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# HIGHLIGHTS OF 2004 BSF SCIENTIFIC/ MEDICAL AND FAMILY CONFERENCE

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Barth syndrome, the speakers were introduced by parents of boys affected by BTHS. The parents provided a brief description of the course of their family's diagnosis, progression of disease and treatment. This served to highlight the disorder's variability of presentation, as well as the frustrations of parents as they search for answers about treatment options and the clinical course of the disease. The scientific portion of the meeting was divided into the basic science presentations on the first day and the clinical aspects of the disorder on the second day.

The age of presentation of Barth syndrome classically has been in the newborn or toddler years when the boys develop signs and symptoms of congestive heart failure or serious bacterial infection. However, the phenotype of BTHS is extraordinarily variable; some patients are severely affected as neonates, while others develop milder symptoms later in life. The determinants of this variability in presentation and clinical course are unknown. Patients with BTHS are hypotonic as neonates and may be neutropenic. They may have evidence of congestive heart failure, prompting initiation of digoxin and afterload reduction. In the toddler years, affected boys have failure to thrive and short stature. The cardiac dysfunction tends to improve or even normalize in the pre-pubertal years. By the teenage years, BTHS patients have a dramatic increase in their growth velocity. At this point, the cardiac dysfunction may recur. Throughout their lifetimes, patients may be affected by serious bacterial infections and recurrent mouth ulcers due to neutropenia. There are on-going issues related to the neurological manifestations of the disorder, such as hypotonia, weakness and myalgia, which

limit the exercise capacity of even those patients with normal cardiac function. Patients with Barth syndrome seem to have normal intelligence and, according to their parents, a "wise" sense of humor.

The basic science portion of the meeting was composed of work on the genetics of the disease and the description of the functional disorder caused by the genetic abnormality. Iris Gonzales, Ph.D. of A.I. DuPont Children's Hospital and

Frederic Vaz, Ph.D. of the Academic Medical Center in the Netherlands presented their work on the genetics of BTHS. The genetic sequence of the TAZ gene predicts multiple potential gene products of TAZ transcription; however, there is one predominant gene product expressed in humans and higher primates. The TAZ gene

encodes tafazzin, which is homologous to members of the acyltransferase superfamily involved in complex lipid metabolism. A patient's particular gene mutation is, however, not predictive of their clinical course. In fact, patients within the same family may be affected differently.

As described by Michael Schlame, M.D. of New York University Medical Center and Miriam Greenberg, Ph.D. of Wayne State University, the primary disorder in BTHS is a defect in the remodeling of cardiolipin, a critical structural component of the mitochondrial inner membrane. Mass spectroscopy performed on human cells as well as various genetic models of tafazzin deficiency show that the mature forms of cardiolipin are not present, and that there is a backlog of cardiolipin precursor products. Tafazzin plays a critical role in the three-dimensional orientation of fatty acid components of cardiolipin. Abnormal

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**"Barth syndrome (BTHS; MIM #302060) is an uncommon, but probably underdiagnosed, X-linked genetic disorder characterized by dilated cardiomyopathy, skeletal myopathy, failure to thrive and neutropenia."**

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tafazzin results in a “stereochemical disorder” of the inner mitochondrial membrane, fundamentally changing the three-dimensional properties of cardiolipin and affecting how cardiolipin interacts with other components of the cell, e.g. proteins and other components of the mitochondrial membrane. This abnormal structure of cardiolipin results in a “leaky” mitochondrial inner membrane. The membrane potential, which provides the driving force for ATP generation, is low; therefore, ATP production may be lower than normal. In order to provide the cell with adequate amounts of ATP, these dysfunctional mitochondria proliferate. There are approximately twice the normal number of mitochondria in affected cells. Work done in yeast

using two mutant strains, one in which there is no cardiolipin produced and one in which the yeast homologue of TAZ is missing, demonstrate that these yeast do not grow in stressful conditions, e.g. high temperature or hypotonicity. Replacement of normal human TAZ in the yeast model corrects the phenotype. The ATP production capacity of the respiratory chain components in an individual mitochondrion from a cell expressing mutated tafazzin is normal, supporting the hypothesis that the primary defect in ATP production is an inability to generate a driving force for ATP production at the membrane level. Arnold W. Strauss, M.D. of Vanderbilt University presented results from his experiments using a

zebra fish model of Barth syndrome. The fish are a “knockdown” model using antisense morpholinos to suppress production of tafazzin protein. The hearts of these fish are abnormally formed and function poorly. They develop large pericardial effusions and bradycardia. Work is progressing on the development of a transgenic mouse model of BTHS. Cardiolipin is also involved in the

mitochondria’s response to low oxygen states, which occurs in ischemia-reperfusion injury, for example. In fact, cardiolipin stores in the mitochondrial inner membrane are depleted during experimental ischemia-reperfusion injury. In addition, the abnormal structure of the mitochondrial inner membrane results in leak of certain mitochondrial components, such as cytochrome c and mitochondrial DNA, into the cytosol. Work by Mauro Degli Esposti, Ph.D. at the University of Manchester showed that there may be a link between the release of cytochrome c and the induction of apoptosis. This may play a role development of neutropenia in BTHS patients.



From left to right: Dr. Randall M. Bryant, Univ. of Florida-Jacksonville/Gainesville; RJ (age 6), held by Dr. Barry Byrne, Shands Children’s Hospital, University of Florida, Gainesville, FL; Dr. Carolyn T. Spencer, Univ. of Florida College of Medicine, Gainesville, FL; and Michael (age 18).

The cardiac manifestations of BTHS include left ventricular non-compaction, dilated cardiomyopathy and, possibly, a propensity to develop ventricular arrhythmia. Infants frequently present with signs and symptoms of congestive heart failure and require treatment, e.g. digoxin and afterload reduction. These medications may be weaned and even stopped as the patient may go through a “honeymoon” period between the ages of 5 and 10 years. The systolic heart function as measured by shortening fraction may be normal during this period. During puberty, there is again worsening of cardiac function followed by improvement in the teenage years. The reasons for this variation in cardiac function

are not known. Carolyn Spencer, M.D. and Barry Byrne, M.D., Ph.D. at the University of Florida, Gainesville are currently conducting a longitudinal study on the cardiac manifestations of BTHS. According to their data, cardiac function as measured by shortening fraction is in the mildly dysfunctional range on average. The wall thickness measurements are normal, as are the chamber

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# HIGHLIGHTS OF 2004 BSF CONFERENCE

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sizes. The progression of the cardiac disease will be followed in their study. It should be noted that this study included only those patients well enough to travel to the study site, so patients experiencing exacerbations of cardiac function may be under-represented.

The propensity of BTHS patients to develop arrhythmias is being addressed as a part of the study at the University of Florida, Gainesville. Randall Bryant, M.D. presented five cases of ventricular arrhythmia and two cases of prophylactic defibrillator implantation in patients with BTHS syndrome. The patients ranged in age from 11 to 21 years. In most of the cases, patients experienced syncope with or without palpitations and had severe ventricular arrhythmia documented either during an event or at subsequent electrophysiological study. In general, BTHS patients with arrhythmia have mild left ventricular dysfunction that has been stable, with a shortening fraction in the 25-30% range. Vasovagal symptoms and chronic fatigue are common among BTHS patients and were present in all of the cases. Three of the patients had male siblings who died with cardiorespiratory symptoms and two of the three had other features of Barth syndrome. Currently, signal average EKGs and microvolt T-wave alternans (MTWA) analyses are being performed on patients with BTHS who are old enough to cooperate with the test. A positive MTWA test has been shown in the adult population with non-ischemic cardiomyopathy to be predictive of adverse events (e.g. sudden death, ventricular tachycardia, ventricular fibrillation) occurring within one year. In general, patients with BTHS should be aggressively evaluated for ventricular arrhythmia as a cause for syncope, palpitations or vasovagal symptoms. Holter monitoring, event monitoring, tilt-table testing and EPS are all appropriate as clinically indicated.

Patients with BTHS have either a congenital or cyclic form of neutropenia according to Colin Steward, B.M., B.Ch., Ph.D. of the Bristol Royal

Hospital for Sick Children. Those with the pattern of congenital neutropenia have an absolute neutrophil count (ANC) less than 500 frequently with mild-to-moderate anemia and a low platelet count. These patients generally experience serious bacterial infections. The cyclic form of neutropenia typically has an oscillating pattern with a 21 day cycle. The ANC nadir is in the 0 to 200 range and can be associated with mouth ulcers. The ANC typically peaks between 1500 and 2100. Some patients with BTHS also have recurrent lymphopenia, especially of the CD8+ subpopulation. It may be difficult to document neutropenia if there is a cyclical pattern, as the cycle duration is not necessarily the typical 21 days. In order to document the pattern of neutropenia, multiple samples may need to be tested over a period of 6 weeks. Patients with recurrent mouth sores or cyclic neutropenia may be treated with GCSF to stimulate neutrophil production. According to some parents, patients may respond to a very small dose of GCSF with an appropriate increase in ANC and resolution of any clinical symptoms.

The neurological aspects of BTHS were highlighted in a presentation by Tyler Reimschisel, M.D. of Johns Hopkins Hospital. Newborns with BTHS are frequently hypotonic at birth while older children have proximal muscle weakness, myalgias and fatigue. In this study, 75% of participants complained of weakness, usually localized to the upper and/or lower extremities. In many patients, prolonged sitting was very tiring which was explained by weakness of the neck, hips, and shoulders on neurological examination. Muscle weakness is not necessarily accompanied by a decrease in muscle bulk, as only one third of patients had subjectively decreased muscle bulk. Myalgias were common, occurring in 71% of patients, especially after a very vigorous day. Headaches were also common among the BTHS population studied, even in those with no family history of headache. Significant headaches occurred greater than once monthly in the majority

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of patients, and one third of patients experienced lightheadedness or felt dizzy. Approximately one third of patients had an abnormal gait (e.g. wide-based, everted ankles or toe walking); nearly half had ankle weakness and three-quarters had a flat arch. The neurological manifestations of the disorder may be quite variable in severity and presentation.

Barth syndrome is a multi-faceted disorder with great variability in course and presentation. The complexity of the disease makes a multi-disciplinary approach to patient care invaluable. Richard I. Kelley, M.D. of Johns Hopkins Hospital and Colin Steward, B.M., B.Ch., Ph.D. discussed the clinic model used at their respective institutions. In either case, the approach to the patient with Barth syndrome involved evaluation by several subspecialists, including cardiology, hematology, neurology, development, metabolism and genetics. For certain patients, evaluation by a gastroenterologist or an endocrinologist may be of assistance. In addition, patients frequently require services from ancillary care providers such as physical therapy, psychology and nutrition and frequent education sessions on the genetic, nutritional and medical aspects of the disorder. The

clinic model at the Bristol Sick Children's Hospital involved the use of play therapists to prepare the children for the more anxiety-provoking studies, such as MRI. In addition, the clinic was "family-led" and addressed the needs identified by the parents. The Bristol clinic is not held "too often" in order to minimize the inconvenience for parents who have to travel long distances to reach the clinic and thus encourage compliance with visits.

Great advances in the understanding of Barth syndrome have been made since it was first described in 1983 by Peter G. Barth, M.D., Ph.D. and colleagues. However, many aspects of the disorder remain to be explained, the more important of which are the factors predicting a more severe course and the link between the genetic defect and the clinical manifestations of the disease. In addition, there is, as yet, no disease-specific treatment for Barth syndrome, a treatment that parents of Barth patients eagerly await. The Barth Syndrome Foundation is helping to move the process of discovery forward by organizing and participating in scientific meetings, funding research and educating parents and physicians about this challenging disorder.



NATIONAL HEALTH COUNCIL

## LATE BREAKING NEWS

December 14, 2004

### BSF Becomes Member of National Health Council

We are pleased to announce that The Barth Syndrome Foundation, Inc. has been accepted as a member of the National Health Council. Acceptance as a member of The National Health Council is considered the gold standard for voluntary health agencies. The Barth Syndrome Foundation is privileged to be in the company of 49 prestigious voluntary health agencies who have been the forerunners in compliance with setting the standards of excellence for voluntary health agencies.

The National Health Council has adopted a set of good operating practices to ensure that its voluntary health agency members maintain the highest standards of organizational effectiveness and public stewardship. The standards cover the areas of governance, personnel policies, programs, fundraising, finance, accounting and reporting, and evaluation. These standards are closely aligned with, but more extensive than, the Standards for Charitable Accountability adopted by the Better Business Bureau's Wise Giving Alliance (WGA). To review the good operating practices of The National Health Council visit [http://www.nationalhealthcouncil.org/aboutus/stand-good\\_operating.htm](http://www.nationalhealthcouncil.org/aboutus/stand-good_operating.htm).

# BSF Board Restructures for the Future

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Why make these changes now, especially as BSF celebrates our most successful year yet? Probably the most important reason is to insure that our success continues. In preparation for our meeting, Gene asked the Board to read the book Good to Great and to consider carefully the lessons learned from studying good organizations that had gone on to become great, as well as those that had stagnated and become undistinguished. We realized that BSF is at a critical point in our evolution. In four short years BSF has:

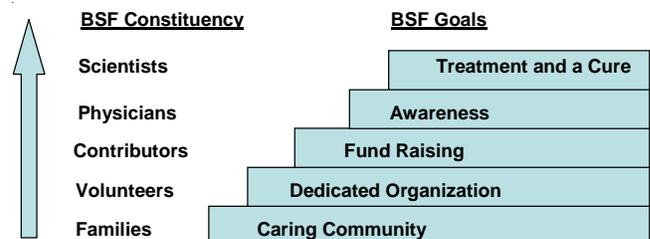
- gathered a critical mass of affected families and successfully built programs that provide them with a broad range of timely informational and educational support
- initiated an international physician awareness program that is reaching more and more physicians and enabling them to recognize and diagnose Barth syndrome cases
- awarded 10 research grants (with an additional six currently under consideration), with the advice and counsel of our world-class Scientific and Medical Advisory Board

Through good luck and good decisions we have made excellent progress in building an informed and caring community for our families. We have begun the task of educating and supporting physicians. These were the first crucial steps that BSF had to take, along with building a dedicated and growing group of volunteers and financial contributors.

Our accomplishments in each of these areas has brought us credibility, experience, contacts and a loyal, growing base of physicians, bench scientists and families whose participation is crucial to the next phase of research into treatments and a cure. Our mission has always been very clear: *To guide the search for a cure, to educate and support*

*physicians and to foster an informed and caring community for affected families. A caring and informed community is at the core of our foundation. We cannot afford to lose our commitment to this goal. Likewise, increasing awareness among physicians is critical if we are to find and save the lives of boys not yet diagnosed. But neither of these two primary goals will cure any of our boys, and frankly, the quality of care for a diagnosed child will improve only marginally, without significant advances in research and understanding. Though we have laid a strong foundation in this area, Science and Medicine must be the next great focus in our strategic plan for the next five+ years.*

## Each Stage of BSF's Success Builds on a Foundation of Earlier Successes



## The Challenge is Daunting.

There are well established foundations and groups which have been working to find a cure for much less rare disorders for years. There are entire labs and experienced scientists dedicated to research into other more prominent diseases. Funding required for these efforts can run well into the millions of dollars. What makes us think that we can raise millions of dollars and attract world class researchers to our cause?

- 1) We have a strong and growing family base. We are consistently admired by other groups because of the growing number of dedicated volunteers who support our cause. *We will never, never, never, never, never give up!*

(Continued on page 11)

- 2) We have already achieved more success, faster than anyone would have believed, other groups are now looking to us as a benchmark. We are becoming the model for others.
- 3) If we don't attempt this, who will? No one is more motivated than we. In short, we have no choice.

We always have a need for resources. The next phase of our work will require the addition of a new set of skills and resources. We need people:

- who can help us attract the attention and support of major research centers;
- who can help us build a stronger bridge to the US NIH (National Institutes for Health), a massive source of research funding, and to other governmental resources in other countries;
- who can help us ally ourselves with other, larger organizations with similar goals so that we can better share resources and enjoy the benefits of scale that we lack;
- who are scientists with insight into the choices we must make in funding research;
- who can help us convince wealthy individuals, charitable institutions and governments to provide the increased funding we will most certainly need.

Finding these new resources and integrating them into our organization is the responsibility of the BSF Board. If we are to be successful in attracting these new resources, the Board itself needs to change. For one thing, given the importance of the advice and contacts and the nature of the decisions we will have to make, BSF would benefit from inviting some of these people to join the Board and help us make these critical decisions.

As successful as we have been, we must look beyond the confines of our own families to find some of these people. There is a concern that inviting non-family members into BSF will change the character of our group, but in fact, we have already done so and been better for it. Our SMAB is comprised of 12 scientists and physicians, none of whom are family members, but all of whom have adopted our cause. Joan Stoner, who is a constant source of educational insight and advice and a regular contributor to the listserv; Jacquie Butera; Eileen Juico; and Jon Rosenshine, who wrote and published "Strategies of Educational Advocacy for a Child Living with Barth Syndrome – An Educator's Handbook," along with "Strategies of Educational Advocacy for a Child Living with Barth Syndrome – A Parent's Handbook," are not family members. Jules Spotts, who advises us on child development; and Gary Rodbell, Tim Monetti and Jon Steigerwald, who have now helped us raise more than \$210,000 through their various Ironman races, are not family members. (Though maybe we should make them all honorary family!)

In appreciation of the need to preserve and reinforce the characteristics that make BSF unique, the Board has proposed that at a minimum, Barth family members must always comprise at least 50% of the BSF Board. Further, any new Board member must be well known to the Board, must be prepared to commit themselves to our values (which accompany this article), our vision and our mission, and accept that they, like we, must never, never, never, never, never give up on our boys. Our by-laws permit up to 21 Board members. We anticipate searching for and adding up to three new Board members over the next year, to better address the critical needs of our organization around the world in the areas of science and medicine, fundraising, and education, and to build closer relationships with other important institutions.

# BSF Board Restructures for the Future

*(Continued from page 11)*

## **The Need for Full Time Leadership**

In addition, as our volunteer leadership strengthens, we acknowledged that it was time to create more of a distinction between the role of the Board and the roles of the program leaders. BSF and our programs are growing quickly and more complicated, and our organization and leadership need to grow and manage those positive changes. Shelley Bowen has organized and led an annual volunteer retreat in Steinhatchee, FL for the last several years. You can read more about that gathering elsewhere in this Newsletter, but this year, reflecting our existing and planned programs, the invited group of BSF program leaders was larger and more dedicated than ever. BSF has now grown to require a full-time, compensated leader. And at this stage in our development, the Board decided that no one could fill that role better than Shelley Bowen. Shelley has consistently demonstrated the vision, the drive, the caring, the leadership skills and the good judgment that has brought us to where we are today. We could not imagine BSF without her leadership, so we offered Shelley the job as full-time President of the Barth Syndrome Foundation, and thankfully, she accepted!

Change is difficult. It is challenging and can be stressful and unsettling. It also can be essential. When measured against the war that might be won and the risk of further loss of our boys, for BSF there can be no alternative. For those whose passion and constant dedication have brought us all this far, we are eternally grateful. Each member of the Board struggled to find his/her best seat on the BSF bus as we stepped up to the new challenges before us. For Anna Dunn and Lynda Sedefian, that decision led to giving up their existing seats to find new ones where their talents and passions can best serve the interests of BSF and the Barth families. For Lynda, that will be as Newsletter Editor and as executive administrative assistant for BSF. Anna has reminded us all to remember why we are here and to what we must always be dedicated, and she also clearly described where her seat, and her heart, are and have always been – in support of the Barth families.

## **Just the Beginning!**

None of us is or should expect to be in our seats forever. When there is someone who can do our job better and with equal passion and dedication, we will always step aside and find new seats. That is how BSF will best move forward. When the challenges have changed and we no longer are the best fit, we will change seats. If a change can make our boys better, we will embrace it. In the end, all of this must keep alive the most important issue of all for all of us... and that is hope for the future.



## **The Values That Guide Us**

- We can make a difference in the lives of those affected by Barth syndrome and we will actively seek to do so.
- We will insure that BSF means: Credibility, Integrity, Professionalism and Compassion.
- We will be accountable for our commitments and actions.
- We will be respectful of the time and talents we are offered and good stewards of the resources we are given.
- We value teamwork and collaboration and constantly seek to improve by learning from others.
- We believe that families and physicians should be able to make their own decisions about care and treatment, and we will help them by insuring access to the latest tools and information.
- When representing BSF, we place the interests of all those affected by Barth syndrome above the interest of any individual.

# Reflections for the NHLBI On the Second Biennial International Scientific/Medical and Family Conference

By Katherine McCurdy, Vice President of Science and Medicine, BSF

Reprinted with permission from NHLBI (FYI from the NHLBI, Volume 5, Issue 2, September 2004)



*Dr. Peter Barth addresses the scientific community on the latest discoveries of Barth syndrome.*

**B**SF would like to report on the great success of its second biennial International Scientific/Medical and Family Conference that was held in Orlando, Florida, in July. Barth syndrome is an X-linked recessive condition that encompasses cardiomyopathy,

neutropenia, skeletal muscle weakness, and growth delay. Because both “heart” and “blood” components are involved in the syndrome, people associated with the NHLBI had a significant presence at the meeting for scientists and physicians. Dr. John Fakunding, Director of the Heart Research Program in the Division of Heart and Vascular Diseases, kicked off the Scientific/Medical meeting with an overview of the NHLBI perspective. Later, Dr. Susan Old, the Associate Director of the Clinical and Molecular Medicine Program, gave a talk entitled “Genetics, Genomics, and Proteomics.” She described the technical resources in these leading-edge areas that are available to researchers through the NHLBI. After having heard Dr. Old speak at the Public Interest Organization meeting in February, we realized that the information she presented was so valuable and timely that we invited her to give essentially the same talk at our conference. She was kind enough to do so. Consequently, she and one of the researchers funded by the Barth Syndrome Foundation now are collaborating to investigate the creation of an antibody that is needed by a number

of scientists working on the disorder. Dr. Liana Harvath, Deputy Director of the Division of Blood Diseases and Resources, also attended our conference, and provided the perspective of the NHLBI blood program, which was wonderful.

One of the other factors that made our meeting so successful was the structure of the conference itself. Our first conference two years ago included two simultaneous tracks of sessions – one for scientists and doctors and the other one for families. At the most recent meeting, a third track was added – one for the affected boys themselves. We find that this framework serves our needs incredibly well.

Having researchers and physicians meet together is a very positive experience. Interaction among researchers and within the physician communities has obvious benefits, but we also see tremendous advantage in having the bench scientists and the clinicians meet together. The basic scientists are intrigued by the clinical manifestations of the processes that they study on a cellular or even molecular level. In turn, the doctors who treat patients with the syndrome often gain a lot from an increased understanding of what occurs on a microscopic level.

I believe that including the families of those affected by Barth syndrome in simultaneous meetings at the same location made the real difference in our conference. Because Barth syndrome is rare (though almost definitely much less rare than commonly thought), most physicians have seen only one case, if any. And, of course, most bench scientists never have any exposure to patients with a condition related to the basic science they study. This convergence of people interested in an

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**“I have always been involved in my particular research because I am fascinated by it and I love working on it, but having come to your meeting, I now know that what I do also is important.”**

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uncommon condition on so many levels is a unique, and often very compelling experience for everyone involved. The physicians are intrigued by seeing additional patients with the syndrome and being able to make even cursory comparisons. Furthermore, we received many comments from basic scientists, such as, “I have always been involved in my particular research because I am fascinated by it and I love working on it, but having come to your meeting, I now know that what I do also is important.”

The patients and their families benefit as well. In this conference model, families have access to the world’s experts in specialties that are very important to them. Because people with Barth syndrome, like those with many other rare genetic disorders, now fortunately are living much longer and, with continuing advances, have a much more promising future, it is really important that the boys and young men with Barth syndrome themselves have an opportunity to meet with these experts as well. During our previous conference, we offered some opportunities for the boys to meet with various world-class physician experts on the disorder. Building on that success, this year, we also held sessions, just for the patients, with basic researchers on subjects such as “I am the Research.” During one such session, the young men affected by Barth syndrome had a chance to talk with and ask hard questions of a bench scientist who studies important aspects of the science related to the condition. The young men thought it was great, and so did the biochemist who led the session.

I think it is safe to say that all those who attended that recent Barth Syndrome International Scientific/Medical and Family Conference came away with a renewed sense of strength, community, and collaboration and a reinvigorated sense of determination, urgency, and commitment. This conference model may not be appropriate for every condition, but in the right circumstances, it can work beautifully.



## **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. (Expires Sept. 2, 2006 unless reissued.)***

### **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. (Expires July 30, 2006 unless reissued.)***

# ANNOUNCEMENT

## COVERAGE OF BARTH SYNDROME FOUNDATION

2004 INTERNATIONAL SCIENTIFIC/  
MEDICAL AND FAMILY CONFERENCE  
July 8-12, 2004

Disney's Coronado Springs Resort  
Lake Buena Vista, Florida

BSF is proud to offer the sale of DVDs covering 20 hours of footage from presentations given by world-renowned experts on Barth syndrome throughout the two days of scientific/medical and family sessions at BSF's 2004 International Scientific/Medical and Family Conference. It also incorporates powerpoint slide presentations given from those experts in the fields of cardiology, hematology, and neurology. In addition, it includes family testimonies highlighting their experience in the historical path of coming to an accurate diagnosis of Barth syndrome. The complete set of DVDs is being offered at a cost of One Hundred Fifty Dollars (US\$150.00). Additional sets are discounted. If interested in purchasing, visit:  
<http://www.finalfocusvideo.com/barth/>

## Save the Date For BSF's 2006 Conference

The Barth Syndrome Foundation is pleased to announce that we will be returning to Disney's Coronado Springs Resort in Lake Buena Vista, Florida on **Monday, July 3rd thru Saturday, July 8th, 2006** to host BSF's biennial International Scientific/Medical and Family Conference. Please mark your calendars and start saving for this important conference.

July 3, 2006	Family Reception
July 4-5, 2006	Clinics
July 5, 2006	Scientific/Medical Reception
July 6, 2006	Family & Scientific Sessions
July 7, 2006	Family & Scientific Sessions
July 8, 2006	Family & Scientific Sessions

If you would like further information, please contact:

Shelley Bowen: [sbowen@barthsyndrome.org](mailto:sbowen@barthsyndrome.org)  
Jan Kugelmann: [jkugelmann@barthsyndrome.org](mailto:jkugelmann@barthsyndrome.org)  
Lynda Sedefian: [lsedefian@barthsyndrome.org](mailto:lsedefian@barthsyndrome.org)

## WWW.BARTHSYNDROME.ORG

By Lynn Elwood, Webmaster, BSF



What's new on the **W**Barth Syndrome Foundation website? On the main page, the *What's New* section has been added. This change was a frequent request from people who visit the site regularly. This will help you to learn what new things have

been posted and find them easily. For those that followed details about the 2004 conference we're hoping it provided you with a chance to easily reach current material and agendas.

Also new are the *Awareness* and *Fundraising* pages. Here we keep a running list of the upcoming conferences the BSF will attend and planned fundraising events. Watch the fundraising page for notes on how to help with events, who to contact and how to contribute. There are other new sections planned for the near future.

The international Barth Syndrome Foundation site is [www.barthsyndrome.org](http://www.barthsyndrome.org).

There are other sites for some parts of the world:

Barth Syndrome Trust (UK& Europe)  
[www.barthsyndrome.org.uk](http://www.barthsyndrome.org.uk)

Barth Syndrome Foundation of Canada  
[www.barthsyndrome.ca](http://www.barthsyndrome.ca)

Barth Syndrome Foundation / South Africa  
[www.barthsyndrome.org/South\\_Africa.html](http://www.barthsyndrome.org/South_Africa.html)

Please send your requests and comments about the website. It will continue to grow and evolve and we'd like your feedback on the directions we should take. You can send feedback to [lwood@barthsyndrome.org](mailto:lwood@barthsyndrome.org) or post to the Listserv if you'd like to discuss it with the group.



# Cardiac Evaluation in Barth Syndrome

By Carolyn T. Spencer, M.D., Barry J. Byrne, M.D., Ph.D., Randall M. Bryant, M.D., University of Florida, Division of Pediatric Cardiology



*Dr. Carolyn Spencer (center), Jan Kugelmann (left), and Julie Berthy, RN (right) review the schedule of the cardiology clinic held during BSF's 2004 conference.*

The natural history of Barth syndrome has not been fully described. In addition, there is significant clinical variability in the cardiac, skeletal muscle, and hematologic phenotype of these patients that is not understood. Even in individual

families the severity of the disease may differ. The faculty at the University of Florida, Division of Pediatric Cardiology has the privilege to be involved with the Barth Syndrome Foundation and in 2004 we collected data on 31 boys (nearly 1/2 of the subjects worldwide known to the BSF). This data collection occurred at the UF Clinical Research Center in Gainesville, FL and at the 2004 BSF conference in Orlando, FL.

The immediate objectives of this project were to collect cardiac, skeletal muscle, hematologic, and biochemical data to better describe the range and type of clinical manifestations of a cross-section of boys with Barth syndrome. The long-term objectives are to collect data in individual boys over time and better understand the changes that occur in individuals with growth and age, and thus learn more about the natural history.

## CLINICAL DATA

**Medications:** Medication history was available in 2004 for 30/31 boys. Of these 30 boys, 19 were on digoxin, 9 on diuretics (lasix/aldactone), 18 on afterload reduction (captopril/enalapril, etc), 10 on

beta-blockers, 7 on GCSF, 7 on Coenzyme Q<sub>10</sub>, 5 on carnitine, 2 on fludrocortisone, and 4 on no medications.

**Growth:** The majority of patients manifested growth deficiency although there was a wide range of growth percentiles for age. For height, 21/31 were < 10% for age, and for weight 23/31 were < 10%. However, the average height percentile was higher in those older than 15 years of age compared to younger than 15 years (29% vs 8%), suggesting that there may be some delayed growth of height during puberty.

**Diagnosis:** Of the 24 boys with a history of cardiomyopathy and available data, all were diagnosed with cardiomyopathy at less than 3 years, and 17/24 at less than 3 months. The average age at diagnosis of Barth syndrome was 3 1/2 years (range prenatal – 15 years). Of 26 patients with available family histories, 11 (46%) have a family history of death either suspected or confirmed to be Barth syndrome.



*Nicholas, of England, cooperates with tech while receiving his echo at BSF's 2004 conference*

## CARDIAC DATA

**Echocardiography:** The average ejection fraction for the 31 boys was 50 ± 10% (range 29 – 67%) and the average shortening fraction was 28 ± 5% (range 13 – 36%), indicating that as a group the cardiac function is low normal,

although there is marked variation among the boys. As a group, the 15 boys for whom echocardiographic data was obtained in both 2002 and 2004 suggest that there was no significant interval worsening of cardiac function or progressive enlargement of the heart.

**Arrhythmia:** Analysis of the ECGs and signal averaged ECGs revealed that many boys had non-

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*Drs. Randy Bryant (left) and Barry Byrne (right), of the University of Florida*

s p e c i f i c abnormalities of the ECG. Of 21 boys who had signal averaged ECG, 3 were abnormal. Of the 17 who had a microvolt T-wave alternans ( T W A )

performed, 2 (already with ICDs) were abnormal. There are currently 7/31 boys with internal defibrillators (ICD), of whom 5 have had documented ventricular or supraventricular arrhythmias or a positive EP study.

#### **LABORATORY DATA**

- 1) Of the boys who had CBC (complete blood count) evaluated, 25% had evidence of neutropenia.
- 2) Prealbumin is a marker of poor nutrition, and 15/19 boys had low prealbumin.
- 3) BNP (brain natriuretic peptide) is a blood marker that is elevated in patients with reduced cardiac function. There were elevations in BNP in many boys evaluated, and this correlated with the ejection fraction.
- 4) Of all labs tested (CBC, lipids/cholesterol, prealbumin, BNP), the only marker that correlated with reduced cardiac function was BNP. There was no evidence that those with neutropenia had a worse ejection fraction.

#### **SUMMARY**

Evaluation of this group of boys with Barth syndrome may be biased toward obtaining data on those who are currently living and doing well enough to travel, thus some of the more fragile boys are not included in this analysis. As we know, there are many boys who have died in infancy and childhood and cannot be included. However, this data

suggests that although the majority of boys have some degree of cardiac dysfunction, they may remain fairly stable over time. Many boys can do well with current medical management strategies. The low prealbumin values also suggest that many boys have some degree of poor nutrition.

The arrhythmia issue is more difficult and was the predominant concern of many parents. Unfortunately, the ability of our current available tests to predict arrhythmia (even in many adult high risk populations) is extremely limited. There were no specific markers in this group of patients that predicted which boys had a proven risk of arrhythmia and have had ICDs placed. The range of reasons that these boys were identified as having a risk of arrhythmia were broad and included abnormalities on screening Holter monitors by their primary cardiologists, syncope (passing out), and positive EP studies performed in the electrophysiology laboratory. Although some boys with arrhythmia risk may have a positive TWA, there have also been cases of negative TWA with documented arrhythmia. So, although this test may be helpful, it does not appear to be the “gold standard”. Abnormal signal averaged electrocardiograms in some of the boys suggest that the causes of arrhythmias might be multifactorial.

In order to develop a better understanding of these issues, it will be imperative to continue to follow these boys over time and to continue to expand the group that are participating. There is no “magic formula” for how often these boys should be evaluated by their primary cardiologist, what medications they should use, or how often certain tests performed. However, at a minimum a yearly echocardiogram, ECG, and Holter monitor would be recommended with enhanced follow-up in boys with reduced cardiac function or symptoms of heart failure, syncope or palpitations.

We look forward to continuing to work with the BSF and the truly wonderful group of boys and their families. Please contact us with questions.



# Cognitive Development Project ~ Children with Barth Syndrome

*Dr. Michele Mazzocco, Principal Investigator, John Hopkins University School of Medicine, Kennedy Krieger Institute*

This project is designed to understand the development of cognitive and academic skills in young children. One component of the project involves following the development of these skills in young children with Barth syndrome. We are currently recruiting children who are either in Kindergarten, First, Second, or Third grade, and who have Barth syndrome.

Participation will involve several hours of psychological and academic achievement testing, over one or two days. The testing will include measures of reading, mathematics, spatial reasoning, and other problem solving skills. Parents may receive a summary of their child's test performance following each evaluation, if requested. The testing will occur at the Kennedy Krieger Institute in Baltimore, Maryland, or elsewhere, depending on your geographic region of residence. **There is no charge to you for any of this testing.**

If you desire more information, or if you wish to enroll your child in this project, please contact Dr. Michele Mazzocco, *Principal Investigator of this research project*, at (443) 923-4125, or Anne Henry, *Research Assistant*, at (443) 923-4121. If you prefer, you may also e-mail us at [henrya@kennedykrieger.org](mailto:henrya@kennedykrieger.org). There are no significant risks to participation, nor any direct medical benefits. Minor risks include finding some of the activities too challenging or too easy.



# Overview of Occupational Therapy Clinics at BSF's 2004 Conference

*Dr. Raghad Schroeder, OTR/L, TheraPlay*



*OT Raghad Schroeder with "R.J." during the OT clinic held at BSF's 2004 conference*

During the few days spent at the Barth Syndrome International Scientific/Medical and Family Conference, Paula Geigle, PT, PhD, NCCAM Postdoctoral Fellow and I spent a great deal of time with the Barth boys and their families learning, bargaining and analyzing.

Within the two-day clinics, we saw a variety of ages from almost two years old to 22 ½ years of age.

Many of the boys had a variety of different experience with physical and occupational therapy. Some of the children had not participated in any therapy intervention, others had some therapy and still others had intensive therapy.

We spent a lot of time having the older boys participate in tasks that tested them in various positions, thus assessing their postural strength, mobility and endurance as well as performing certain tasks to assess their fine motor skills. In the younger boys, we performed various activities on the floor to test the same postural and developmental skills. We talked to the boys and their families about their lifestyles, the limitation that Barth imposes on their ability to perform activities such as playing with their friends, finishing a day at school, participating in exciting outings,

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etc. Our overall “informal” findings are summarized as follows:

- The earlier that physical or occupational therapy was instituted the more solid the boys looked especially within the group of boys’ ages 10 to 22 ½ years. This was due to the institution of some sort of “exercise” program as well as instituting strategies for postural support including orthotic inserts. Energy conservation techniques were taught to the family, the child and the supporting services including school, babysitter, teacher, etc., increasing their pacing and endurance.
- We preached the use of “exercise” to tolerance without pushing the boys over the edge. We spent many conversations explaining the difference between anaerobic [short intervals of exercise spread out over a period of time] and aerobic exercise [longer term escalation of activity, increasing the frequency slowly and then going to a short cool down]. The anaerobic exercise, we felt from our limited experience, would be more cardiac friendly. Paula addressed this in length at her presentation to the families.
- The use of Orthotics, especially for the feet, helped with shoulder, spinal and pelvic alignment, thus preserving the bone structure from the effect of low muscle tone. Orthotics also increase the endurance of walking as they align the foot to more effectively use the muscles.
- We spent a lot of time encouraging the younger boys to stay active and become involved in activities that improve cardiac status and physical endurance without undue stress on the heart. Swimming was a favorite of both Paula and I. The martial arts were also seen as a good balance of activity without the contact

sport or the competition with peers. With the older boys, we negotiated, often encouraging them to try a physical activity for a short period of time gradually increasing repetitions or resistance. Often times we see that chronically ill children become “couch potatoes” especially with all electronic opportunities available to entertain them. A recommendation of balances between movement activity and sedentary activity was a common theme with the teenagers. The activity level should, of course, be modified to meet their health needs.

In general, we saw more gross motor issues than fine motor issues. Many of the fine motor issues of writing and cutting in the school day were directly related to poor posture strength, alignment and endurance. We recommended many foot Orthotics, mostly UCB’s that fit inside the shoe.

I heard many stories about eating/swallowing/nutrition issues surrounding the Barth boys. There were many similarities and basic concerns regarding lack of good nutrition [pickles are salty but not full of calories!]. There were a wide variety of approaches to deal with these eating issues depending on the health of the child and the severity of the problem. In general, the eating repertoire seemed to improve with age but at a great cost emotionally, as well as physically, to all involved. I would like to suggest a feeding clinic in the upcoming conference. I know I would love to participate in this venue that is near and dear to my heart.

In closure, I would like to say that with all the advice we offered, both Paula and I agreed that we took in more information than we shared. Allow me to thank all the parents for sharing their boys, their stories, their triumphs and tribulations. I am in awe at the power of these parents and the achievements that this foundation has attained in such a short period of time. I am looking forward to seeing everyone again in two years.



# RESEARCH UPDATE :

## A MOUSE GENE ABLATION MODEL OF BARTH SYNDROME

By Arnold W. Strauss, M.D., Professor and Chairman of Pediatrics, Vanderbilt University



For many years, my laboratory has studied the genetic causes of pediatric heart diseases, especially those causing cardiomyopathy, heart muscle dysfunction, related to mitochondrial energy production. **Mitochondria** are the organelles, or parts, within all

cells that are essential for energy generation. Because the heart consumes huge amounts of energy to maintain its continuous pumping function, energy production in heart mitochondria must be highly efficient. In the normal heart, this energy is generated by the breakdown of fat in mitochondria, a complex, multi-step process that requires over 20 genes. We have examined genetic disorders of this fatty acid breakdown pathway in mitochondria for over 20 years and shown that these genetic diseases cause cardiomyopathy, sudden death, heart arrhythmias, and skeletal muscle dysfunction. These are all also common manifestations (also called the phenotype) of Barth syndrome. In our studies, we created animal models of fatty acid oxidation disorders by deleting these genes from mice. We showed that these genetically-altered mice had the phenotype of human patients with mutations in two fatty acid oxidation genes, very long chain acyl-CoA dehydrogenase and trifunctional protein, key enzymes in breaking down fat. These animal models of fatty acid oxidation disorders have been very useful in understanding why deficiency causes the phenotypes, how the gene deficiency alters energy generation, how the mouse changes (a process we have called adaptation) in response to the gene deficiency in an effort to survive, how this adaptation can be detrimental to the mouse (such as causing obesity and tumors later in the mouse lifespan), and how to treat the genetically-altered mice to avoid sudden death and heart muscle dysfunction. That is, these

animal models, which allow us to study literally thousands of animals, are critical in understanding the human fatty acid oxidation gene deficiencies.

Following the discoveries that Barth syndrome was caused by mutations in the tafazzin (TAZ) gene; that the mitochondrial phospholipid, **cardiolipin**, was deficient in Barth syndrome patients; and that TAZ is homologous to fatty acid (also known as acyl-) transferases, proteins that move fatty acids from one type of fat or phospholipid to another, it was clear that Barth syndrome was likely a disorder of mitochondrial function, our specific area of interest. Cardiolipin (literally “heart fat”) is very abundant in heart mitochondria, functioning as a sort of “glue” to hold the many proteins necessary to make energy together and is essential for mitochondrial function. However, it was apparent that the how and why (that is, the mechanisms) by which TAZ deficiency altered cardiolipin and mitochondrial function were unclear. We knew that, because of the difficulties of doing extensive experiments in boys with Barth syndrome, the creation of an animal model of Barth syndrome would be essential to study these mechanisms. We also believed that any experiments to test ways to overcome or modify the problems caused by TAZ deficiency in humans could be performed much more readily, safely, and quickly if we could do this in an animal with TAZ deficiency.

We knew that doing such experiments to both create and study the phenotype of an animal model of a genetic disorder is expensive and can take several years, so that funding from the National Institutes of Health, the primary source of biomedical research dollars in the United States, would be needed. However, getting grants from NIH is highly competitive, so we knew that generation of some preliminary results to prove the feasibility of creating such animal models would be needed. For this reason, and because of the focus of the

*(Continued on page 21)*

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Barth Syndrome Foundation, we applied for a grant from BSF to create a mouse TAZ gene knockout, a genetically-altered mouse missing the TAZ gene, just as in boys with Barth syndrome. We also knew that our studies required that we look at when and where TAZ is made in various organs of the body.

The simplest method to study where and when TAZ is expressed is by having an antibody to TAZ. Despite the fact that TAZ deficiency has been known to be the cause of Barth syndrome for almost 10 years, no reliable antibody to TAZ has been made, so no one has been able to find out when and where TAZ exists within our organs and cells. Antibodies are made in response to anything foreign. We make them in response to viral and bacterial infections or to immunizations, and these antibodies protect us from such infections. To make an antibody, we compared the amino acid (amino acids are the building blocks of proteins like TAZ) sequence of TAZ from mice, humans, rats, and several other organisms that we found in databases derived from the human and animal genome projects. Although TAZ is very similar in all mammals, there are some amino acid differences. We took advantage of this to design a way to generate an antibody to mouse TAZ. We tested to see if this antibody was specific (i.e. reacted only with TAZ and NOT with other proteins) for TAZ, which it was. Using this antibody, we have shown that TAZ is very abundant in heart, muscle, liver, and brain, organs that do have lots of mitochondria and high energy requirements. This is an important advance, as this antibody is crucial to precisely locating where and how much TAZ is present in both the animal models and in humans, including boys and women who are carriers for Barth syndrome and TAZ mutations. The availability of funds from the BSF was essential to create this much-needed antibody.

To delete the TAZ gene in mice and create a mouse model of Barth syndrome is complex and requires many months. We are in the process of doing that study and hope to have such a model within 6-12

months. Because mice are surprisingly similar to humans, deletion of the TAZ gene in mice should produce a very similar set of abnormalities to those found in Barth syndrome patients.

However, we also discovered that TAZ is made in zebrafish. These pretty, striped fish that are often found in home fish tanks develop very quickly (about 72 hours from egg to swimming fish) and genetic manipulation is very simple. So, we knocked out the zebrafish TAZ gene right after the start of embryonic life in hundreds of fish. We showed that the developing fish have a very abnormal heart, poor development of the tail muscles, and symptoms of heart failure, somewhat similar to boys with Barth syndrome. We also showed that we could rescue the abnormal fish by simultaneously adding back normal TAZ, resulting in normally developing fish. By using a mutant TAZ, similar to one of the human mutations found in Barth patients (G197R), we could show that the mutant TAZ partially rescued the fish, but not completely. In this way, we will be able to study various Barth mutations in this model of the disease. Again, the BSF grant received by our laboratory paid for this research and has been essential to its success.

Dr. Khuchua and I attended BSF's 2004 International Scientific/Medical and Family Conference in Orlando in July. We were delighted to meet other scientists working on various aspects of TAZ and cardiolipin. We learned a great deal about the field. We also gained an appreciation of how little is actually known about TAZ, especially as to its precise function. From the scientific results presented, it is apparent that our research will address many key questions that remain. Just as importantly, however, by talking with and hearing the stories of families with Barth patients, we gained a much broader understanding of the disorder and its long-term effects. These observations tell us what we need to study in the zebrafish and mice that are TAZ deficient.



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- Gilbert-Barness E. **Metabolic Cardiomyopathy and Conduction System Defects in Children. (Review).** *Ann Clin Lab Sci.* 2004 Winter;34(1):15-34.
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- Kenton AB, Sanchez X, Coveler KJ, Makar KA, Jimenez S, Ichida F, Murphy RT, Elliott PM, McKenna W, Bowles NE, Towbin JA, Bowles KR. **Isolated left ventricular noncompaction is rarely caused by mutations in G4.5, alpha-dystrobrevin and FK Binding Protein-12.** *Mol Genet Metab* 2004 Jun; 82: 162-6.
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- Lee DS, Mamdani MM, Austin PC, Gong Y, Liu PP, Rouleau JL, Tu JV. **Trends in heart failure outcomes and pharmacotherapy: 1992 to 2000.** *Am J Med* 2004 May 1;116(9):581-589.

- Cristea IM, Degli Esposti M. **Membrane lipids and cell death: an overview.** *Chem Phys Lipids.* 2004 May;129(2):133-60.
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- Hatch GM. **Cell biology of cardiac mitochondrial phospholipids.** *Biochem Cell Biol.* 2004 Feb;82(1):99-112.
- Doolan A, Langlois N, Semsarian C. **Causes of sudden cardiac death in young Australians.** *MJA* 2004; 180 (3): 110-112.

*Editor's note: For a complete bibliography on articles relevant to Barth syndrome, please visit our website at [www.barthsyndrome.org](http://www.barthsyndrome.org)*

## PRESENTATIONS/POSTERS GIVEN ABOUT BARTH SYNDROME IN 2004

- Katherine R. McCurdy; "A family's experience with a rare disorder: Barth syndrome"; Issues in Genetic Counseling 3 course for second year students in Human Genetic Graduate Program at Sarah Lawrence College, Bronxville, NY; November 29, 2004.
- Zaza Khuchua, Zou Yue, Arnold W. Strauss; "Zebrafish (*Danio rerio*) model of Barth syndrome"; American Heart Association Meeting; New Orleans, LA; November 7, 2004.
- R. A. Newbury-Ecob, R. P. Martin, A. M. Hayes, M. M. Williams, L. A. Tyfield, C. G. Steward; "Barth syndrome (x-linked cardiac and skeletal myopathy, neutropenia and organic aciduria); rarely recognised, frequently fatal"; American Society of Human Genetics Conference; Toronto, Canada; October 30, 2004.
- Iris Gonzalez; "TAZ mRNAs in Barth syndrome subjects, alternative splicing and exon evolution" (poster); American Society of Human Genetics Conference; Toronto, Canada; October 26-30, 2004.
- Uta Schaefer, Adam Hurlstone, Mauro Degli Esposti; "The mitochondria-dependent death of neurosensory cells *in vivo*" (poster); Glasgow, Scotland; Fall 2004.
- Mauro Degli Esposti; "Swimming to death: a sniff into developmental apoptosis"; European Cell Death Organization, 12<sup>th</sup> Conference on Apoptosis; Chania, Crete, Greece; September 18, 2004.

*Editor's note: We are sure that these are not the only presentations made about Barth syndrome during the year, but they represent a sampling from the last few months. Please let us know when a presentation is made so that we can include it in this list.*

# A BSF Thanksgiving

*By Stephen McCurdy, Chief Financial Officer, BSF*

In every culture there are times and ways to count one's blessings - to appreciate the joys of family, of friends and of life. An American secular tradition of which I am particularly fond is Thanksgiving - a time for all of us who find our lives a bit too hectic to pause for a moment and reflect upon our good fortune – and an especially apt time for me to be writing about fundraising for BSF!

There are two things that are common to every program run by BSF – from Research Grants to Family Support, from Awareness to Volunteer Development. What are they? It's simple: Every program is an essential element of finding, caring for and someday curing our boys... and every program takes money to operate. BSF has been able to award over US\$173,000 in research grants in 2004, hold a multi-track International Scientific/Medical and Family Conference, publish two Newsletters, create affiliates in the UK, Canada and South Africa, attend medical conferences to get the word out about Barth syndrome, and run websites and Listservs for families and scientists/physicians! Our budget for 2004 amounts to more than \$400,000, including research awards and our biennial International Conference. And for all of this, we must depend on the kindness of our families, our friends, and sometimes even on strangers.

Thankfully, we have a growing group of families and good friends who realize how important fundraising is and have taken it upon themselves to help raise money for BSF. Before the year's end, I expect that we will have raised over \$220,000 from a series of grass roots efforts, far surpassing similar efforts in any previous year. These friends are daily proof that you do not have to be a professional fundraiser to be able to help BSF, just a devoted friend!



*Gary Rodbell, Will McCurdy, Tim Monetti, John Wilkins and Jon Steigerwald come together to support BSF at the 2004 Wisconsin Ironman Triathlon*

## **2004 Wisconsin Ironman**

Our single biggest fundraising event centered on the 2004 Wisconsin Ironman Triathlon which was held in Madison, WI on September 12<sup>th</sup>. Gary Rodbell, Jon Steigerwald and Tim Monetti each raced over 140 miles in a single day and raised over \$151,000 for BSF! More than \$1,000 for each mile! Gary, Jon and Tim joined with a number of BSF families to write letters to their friends and family members telling them about Barth syndrome, BSF and their effort to raise money for our cause. Letter writing campaigns are often the simplest and best way to raise money. They are very low cost and when written as a personal letter from friend to friends, make a very effective case. For this Ironman, donations were received from 357 individuals and 12 companies with matching gift programs. Donations came from contributors in the US, Canada, Japan, the Philippines, the United Kingdom and Sri Lanka. The largest six donations ranged from \$5,000 to \$13,000, but the average of the rest was just over \$250. As a result of everyone's efforts, BSF won second place in the

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(Continued from page 24)

Janus Charity Challenge, which encourages triathletes to use the Ironman to raise money for their favorite cause. In recognition of our success, Janus contributed a bonus of \$8,000 to BSF.

Steve and Will McCurdy, along with Mike, Sue and John Wilkins, went to Madison to cheer on our Ironmen. All three men finished this grueling race and then thanked us for coming! Gary, Tim and Jon – the real thanks goes out from all of us to you. You have shown us all that your hearts are large enough to complete an Ironman race and to carry the hopes of a hundred boys on your shoulders at the same time.

### Third Annual BSF Golf Tournament



Despite three hurricanes and two postponements, Jan and Steve Kugelmann successfully held their Third Annual BSF Merritt Island Golf Tournament. One Hundred and Forty Four (144) golfers (and near-golfers) lined up to tee off on a gorgeous sun-drenched Florida day on The Savannahs golf course on October 3<sup>rd</sup>. Sixty nine businesses and individual hole sponsor names appeared on signs at each tee and were advertised in the event program. In attendance was R.J. Kugelmann's cardiologist, Dr. Barry Byrne (BSF Scientific and Medical Advisory Board member) from the University of Florida, along with his son who left Gainesville at 4AM for the drive to Merritt Island in order to be ready for the 8:30AM shotgun start.

Following 18 grueling holes, longest drive, closest to the pin and putting contests, the many participants on hand joined the Kugelmanns, Dr. Byrne, Shelley Bowen (BSF President) and all the volunteers at the famous Kings Duck Inn for an awards luncheon. Prizes were awarded, and raffle tickets were drawn for gifts donated by local businesses. R.J. Kugelmann drew the winning 50/50 ticket, and we owe a special thanks to John Sherrill who unselfishly gave his windfall back to BSF! Jan and Steve broke last year's record by raising over \$20,000 for BSF this year. The reputation of the BSF Golf Tournament is growing with each passing year, and many of the athletes who took part in this year's fundraiser have already begun to negotiate for an entry into next year's tournament. Tee off slots for 2005 are going fast... better sign up soon!

### Sports Auction

Ed Nottle, Manager of the Brockton Rox, a minor league baseball team in Brockton, Massachusetts, raised \$15,000 for BSF. Ed is an old friend of Tom and Laurie Monahan and a great admirer of their Barth boy, Timmy. Working with Tom and Laurie, Ed organized an evening at Joe Angelo's Pub in Brockton on July 7th, sold raffle tickets for an autographed World Series Champion Red Sox Pitcher Curt Schilling glove, and then auctioned off other autographed sports memorabilia. Especially entertaining was the bidding to shave the head of a local city councilman, Mike Brady. With the active help and encouragement of the Pub's patrons, the group collectively outbid Mike's girlfriend and paid \$1,900 to see the politician's head shaved. Timmy Monahan was given the honor of wielding the razor. Now that is a dedicated and courageous public servant!

Providing support for the Monahans and representing BSF were Anna and Mark Dunn and Shelley Bowen. Shelley also was interviewed on the local radio station and helped increase awareness of Barth syndrome beyond those in attendance. Ed Nottle presented the proceeds from

(Continued on page 29)



*1st Row: Michael, Ashley, Derek  
2nd Row: Steven, Timmy, Jack  
3rd Row: Adam, Ben, Jake  
4th Row: Ryan, Darryl and Jamal, Robert*



*1st Row: Tur, Michael, R.J.  
2nd Row: Nicholas, Colin, Dillon  
3rd Row: Christian, Will, Aldo  
4th Row: Lyem, Kai, John*



*Top Left: Kevin Baffa*

*Top Center: Michael and Nickolas form an everlasting relationship while at the Texas Outreach in Brennan, TX*

*Top Right: Oliver*

*Bottom Left: Jef*

*Bottom Right: Benjamin*



*The photos shown on pages 26 thru 28 are those Barth boys and young men who attended BSF's 2004 International Scientific/Medical and Family Conference. Special thanks goes out to photographer Kelly LaDuke who generously gave her time throughout the conference to capture these special individuals !!*

# Raising the Bar in BSF Awareness

*By Stephen Kugelmann, Vice President, Awareness, BSF*

One of the most rewarding jobs that I have ever been given is to head up the awareness campaign for BSF. To know that you have been personally involved in the proper diagnosis of a child with this disorder is extremely gratifying. The Awareness Team has worked hard to provide critical information to both the medical community and the lay population. I am not sure if it is our charm or our ever enduring persistence that has enabled the group to be so successful. It is with great pleasure that I report that we have made tremendous progress in this arena through multiple mechanisms.

This newsletter, edited by Lynda Sedefian, is one of our most comprehensive outlets for information to all sectors. In its third year of publication it has been well received by all groups. Its distribution has grown to more than 3,000 physicians, contributors and family members. Another area of significant importance is the BSF website managed by Lynn Elwood. Throughout the past year the website has been revamped, including the translating of the opening page into 14 different languages. This was a huge undertaking and one would imagine required multiple volunteers around the world.

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# A BSF Thanksgiving



*Timmy and Aldo  
prepare to throw out the first pitch  
at the "Rox" baseball game*

*(Continued from page 25)*

the night's fundraising to Tom and Laurie at the following Brockton Rox home game. At this writing, Ed Nottle has launched yet another fundraising effort for BSF, raffling off an all expense paid trip to Florida to attend spring training with his team and to visit the World Champion Boston Red Sox and Curt Schilling at their spring training camp nearby. For fans of the Brockton Rox and the Boston Red Sox, this is a dream come true. And for Tom and Laurie Monahan and BSF, Ed Nottle is a dream come true. Thanks, Ed, from all of us!

## **Bowling Night**

John and Liz Higgins sponsored a bowling tournament for BSF in Highland Lakes, New Jersey. Proving that fund raising does not have to be elaborate or difficult, they had a night of great fun with their friends and raised \$1,500. BSF benefits greatly from fundraisers large and small. Each one creates awareness and a greater level of understanding of Barth syndrome. As a result of John and Liz's efforts, Highland Lakes has now heard of Barth syndrome and a child's life may well be saved.

## **BSF Conference and Educational Brochure**

The Wilkins, Bowen, Fairchild, Mann, Kugelmann, Pagano and Sedefian families, as well as other members of the BSF Conference Committee approached a number of individuals, companies and foundations in their local communities and

raised over \$40,000 for the 2004 Barth Syndrome International Scientific/Medical and Family Conference. Special thanks go to Miss Christina Hixson and the Lied Foundation Trust for their lead sponsorship.

BSF and our families are grateful as well to Paul and Allene Russell, Will McCurdy's grandparents, for a special grant which funded the development of two educational handbooks, written by Jon Rosenshine. Jon met with and studied the special educational needs of Barth boys that often result from their fatigue and frequent absences due to illness. Both handbooks were recently published and distributed to BSF Families and their school systems, who have acclaimed it a rich source of guidance where none had previously existed. Thank you Paul, Allene and Jon!

The names of everyone who has donated in excess of \$50 appear on the inside back pages of this Newsletter, along with the names of many friends who have given their time, their expertise or in-kind donations to BSF so far in 2004. I would also especially like to thank all of the brave souls mentioned above who have led fundraising events great and small for BSF. You have proven that fund raising can be fun, and a great generator of awareness for this rare disorder, in addition to a source of much needed funds. To all of our contributors and fundraisers – Thank You! We are all so much better off for your efforts!



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

## Eliza and Tessa's Holiday Fund Raiser



*Eliza and Tessa sell hot chocolate and chocolate chip cookies in Greenwich to raise funds for BSF!*

Was it the secret recipe for the chocolate chip cookies? Or was it the two 10-year-old girls in their blue BSF t-shirts? Or was it location, location, location? Whatever their secret, Eliza McCurdy (a Barth sibling) and her

best friend Tessa Fox successfully raised \$454 for BSF by selling hot chocolate and homemade chocolate chip cookies to holiday shoppers outside Steven B. Fox's Greenwich, CT jewelry store. Eliza and Tessa stopped every passer-by, made their "pitch" briefly explaining the disorder, passed out a brochure (and cookies and hot chocolate) and won a donation. No one could resist! Their three-hour proceeds were then generously matched by Tessa's parents, Steven and Linda Fox, increasing their total to \$908. "Tessa and I wanted to do something for my brother and give BSF a present for the holidays," said Eliza. "It was easy and fun, and besides, it was worth it. Mr. Fox's cookies were really good!"

### HAVE YOU INCLUDED BSF IN YOUR WILL?

One of the best ways to support the continued efforts of the Barth Syndrome Foundation is to remember the foundation in your estate planning. Most folks don't think about this, but it's a great (and easy) way for us to support something we feel strongly about!

By including BSF in your will, you ensure that this good work will continue for generations to come. Talk to your lawyer or estate planning professional about including BSF in your will ... and help to give the foundation the long-term financial base that it needs!

More to come on "planned giving" in the near future ...

## 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 *TAZI*) 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 boys 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 boys at an increased risk of serious infections.

**Muscle Weakness and General Fatigue:** All muscles in a Barth patient, 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 diagnosis is key to survival for Barth syndrome boys.** Those in whom the diagnosis of Barth syndrome is missed have only a 30% chance of living through the first few years of life. With a proper diagnosis at an early age, however, these boys have an 85-90% chance of survival. This is why awareness of Barth syndrome is so important.



*The Cardiomyopathy and Heart Failure Program at the  
Children's Hospital of Philadelphia*

*Announces*

*Barth Syndrome Multidisciplinary Clinics:*

*\* January 13, 2005 \* April 14, 2005 \* July 14, 2005*

*Interested families should contact Genotra Byus, Program  
Administrator at (215) 590-6051, for more information.*

 The Children's Hospital of Philadelphia  
*A pediatric healthcare network*

# Raising the Bar in BSF Awareness

*(Continued from page 28)*

Once again BSF has been present at a host of medical conferences. These conferences have allowed us to connect on a professional level and a personal level with those in attendance. The conferences attended in 2004 were:

- The 7<sup>th</sup> Annual Update on Pediatric Cardiovascular Disease hosted by Children's Hospital of Philadelphia in Orlando, FL attended by Shelley Bowen and Matthew Croxton in February 2004
- American College of Medical Genetics in Kissimmee, FL attended by Jan and Steve Kugelmann in March 2004
- Sudden Arrhythmia Death Syndrome Conference in Salt Lake City, UT attended by Casie Oldewage and Shelley Bowen in August 2004
- Child Neurology Society in Ottawa, CA attended by Cathy Ritter, Lynn Elwood and Shelley Bowen in October 2004
- American Heart Association in New Orleans, LA attended by Mike, Sue and John Wilkins in November 2004
- The Fifth International Symposium on Pediatric Cardiac Intensive Care in Miami, FL attended by Steve Kugelmann and Steve McCurdy in December 2004

Additionally, many presentations in different forums have been given by BSF volunteers to medical organizations and affiliations. Even grade school classrooms were exposed to the key elements for a proper diagnosis of Barth syndrome. Liz Higgins and Michaela Damin have each presented to the local Rotary Clubs here in the U.S. and the U.K, an excellent venue for educating those within the local community about Barth syndrome, BSF and one's personal involvement with this disorder.

Lois Galbraith and Jeannette Thorpe, in Canada and South Africa respectively, have been instrumental in ascertaining key contacts at medical institutions. These contacts have agreed to share our printed information via internal mail and the

"What is Barth Syndrome?" presentation electronically via email with their colleagues. Additionally, Jeannette's persistence was rewarded by the organizer of the Heart Association National Congress held this October in Durban, South Africa (for more information, please refer to page 43). Joke van Loo and Michaela Damin were able to have our scientific brochures on display at the Society of Inborn Errors in Metabolism conference held in Europe. Joke also attended a Dutch mitochondrial diseases' meeting speaking with physicians and families about Barth syndrome.

Our first-ever BSF Outreach meeting, held in Texas, was planned and executed by Shelley Bowen. Geared primarily towards empowering family members to participate in BSF, physicians also were in attendance to share their views on Barth syndrome. Again while travelling to a fundraising event with the Dunn and the Monahan families, Shelley visited the Barbara Bush Children's Hospital at the Maine Medical Center. There she met with the head of the department in pediatric cardiology. Likewise, Lynn Elwood set up a visit to the Chicago Children's Hospital at the University of Chicago. Both were welcomed and once again raised awareness in these geographical areas.

All of the above has played a key role in raising the bar in BSF awareness. Within one year's time, we have delivered to targeted audiences more than 3,000 newsletters. Additionally, over 6,000 scientific and family brochures have been dispersed through these methods. Hundreds of copies of Dr. Kelley's "Diagnostic Criteria and Testing for Barth Syndrome," along with the list of approved labs, have also been distributed. Clearly, it is a significant expense in printing costs. However, it is well worth the cost if we can reach the physician or family of an undiagnosed Barth child.

You know you are doing a meaningful task when the physicians commend you on your involvement in raising the awareness levels of this rare disorder. This statement has been made at every conference that we have attended. But, clearly there is still a

*(Continued on page 32)*

significant amount of work to be done so we can achieve our goal of creating "... A world in which no one will suffer or perish from Barth syndrome.



**Come see BSF at these upcoming conferences in 2005:**

- Eighth Annual Update on Pediatric Cardiovascular Disease in Orlando, FL
- 2005 ACMG Annual Clinical Genetics Meeting in Dallas, TX

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## BEING PROACTIVE HAS MANY REWARDS FOR THIS YOUNG MAN

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*Dr. John Cheatham, Director of Cardiac Catheterization and Interventional Therapy at the Children's Heart Center and Professor of Pediatrics at Ohio State University in Columbus, OH, and Jenny Strawn, RN, BSN, Pediatric Cardiology nurse at the University of Nebraska Medical Center and Children's Hospital in Omaha, NE, stop by BSF's booth to visit with John Wilkins, BSF's representative at the American Heart Association's scientific conference.*

**By John Wilkins (Age 22)**

Recently, as a representative of BSF, I attended the American Heart Association Scientific Conference in New Orleans. My parents and I manned the BSF booth.

It was very interesting to see all the doctors and the representatives from the different medical companies that were present in the exhibit hall. One of the doctors we saw was my former pediatric cardiologist, Dr. John Cheatham, and my former pediatric cardiology nurse, Jenny Strawn (photo above). While there, we spoke to doctors and companies about Barth syndrome. We also saw several of the doctors who are involved with research in Barth syndrome and treating Barth boys. We met doctors who had never heard of Barth syndrome, who gladly listened to us and took our information.

I feel that getting involved in BSF is something that is important for those of us who have Barth syndrome. It is time for us to help raise awareness for Barth so a cure can be found.

What are some of the ways we can do this you might ask? One is to be informed about the symptoms and effects of our disorder. Information about this can be gained via the BSF website, the BSF brochures, or to join the Barth Boys' Listserv (contact Shelia Mann for information on how to do this at [smann@barthsyndrome.org](mailto:smann@barthsyndrome.org)). I always carry our BSF business cards in my wallet and wear a BSF pin. When someone asks about the disorder, I can tell them about Barth, give him or her a card and ask them to visit our website. Another way to get involved is to express your opinion when asked about which direction the foundation should be going. Please feel free to contact Shelley Bowen or put any ideas on the Barth Boys' Listserv and we will get them to her, plus, that way we can talk about them.

I think that as we have the time and the energy available, it would be a good idea to start getting more involved with the foundation. We are the future leaders of the organization.



# BSF HOSTS 3RD ANNUAL VOLUNTEER ENRICHMENT WORKSHOP



**Top Row** (left to right): Steve Kugelmann, Terry & Dick Dannels, Michael Bowen, Michael Telles, Gene Early

**2nd Row** (left to right): Scott Oldewage, Lynda Sedefian, Karen Gordon, Kate McCurdy

**3rd Row** (left to right): Casie Oldewage, Lynn Elwood, Maria Olson, Jan Kugelmann, Sue Wilkins, Susan Dickinson, Susan Hone

**4th Row** (left to right): Shelley Bowen, Shelia Mann, Jess Weiderspan, Steve McCurdy

**Bottom Row** (left to right): Cathy Ritter, Chris Hope, Michaela Damin, Lois Galbraith, Michelle Telles, Jeannette Thorpe

*By Terry & Dick Dannels (Wyckoff, New Jersey)*

I want to take this opportunity to share some thoughts as grandparents attending the Steinhatchee weekend. In a word, the experience was AWESOME, that is, we were awe struck throughout the weekend sharing it with this sincere, caring, and highly energized group. The Board and its helpers had planned the meeting with perfection, from the well-prepared and succinct presentations in group meetings to the roasted croutons for the Caesar salad. We won't mention the grit-balls.

Our interpretation of the underlying principles guiding the retreat were: we share a common bond that makes us family; as family we can speak to one another honestly with love; we will work unceasingly toward our common vision, guided by our mission: *"Saving lives through education,*

*advances in treatment, and pursuit of a cure for Barth syndrome;"* and to this end ... we cannot waste time.

Dick and I were honored to be included in the Steinhatchee process. We love our grandchildren more than we can say. Jack is an inspiration the way he meets the challenges in his life with humor and determination. We are proud of the way Liz and John have fostered this in him.

We came away from the meeting recognizing ourselves as an integral part of BSF. Our commitment to that realization is to periodically evaluate our abilities, gifts and talents and determine where our responsibility lies. For now, here it is:

We will not say, "What can I do?" We will say, "I can and will do...". We will contribute financially each month to BSF -- something, even though a pittance. We learned that if our foundation can show 100% participation in contributions we will more likely be eligible for government and other funding. We will enlist our extended family's support. The more soldiers in our ranks the more likely we can win the war sooner. ***We will win in the end!*** We will host at least one fundraiser a year. Maybe just a candle or a pots and pans party, but something.

We will take advantage of every opportunity to acquaint others with Barth syndrome and its Foundation by name: the medical conferences we attend, at visits to doctors, the bridge clubs, the church meetings, etc. We never know where it can make a difference. We will network with other organizations to promote our goals and foster our mission. We will answer the questions posted on the Listserv in a timely manner and complete the registry as best we can. I have signed up on the Science and Medicine Committee and Dick is on the Awareness Committee.

With thanks to all of you for all you do for all of us.



# WORKSHOP RECRUITS NEW VOLUNTEERS TRULY DEDICATED TO OUR CAUSE

By Casie and Scott (Utah)



*Scott and Casie with their beautiful children Lexie and Christian*

As many of you know, a weekend of intense workshops was held in Steinhatchee, FL from November 19th thru the 22nd, 2004. Twenty-seven people gathered together to examine ourselves and our commitment to this incredible foundation.

Prior to attending this workshop, each individual was responsible to read Good to Great written by Jim Collins. Shelley listed this book as the Book of

the Month on the Listserv. The major goal of this workshop was to make sure that we had the right people on the bus as well as the right people in the right seats on the bus (an analogy used by Jim Collins in the book Good to Great).

We also singled out what we are most passionate about, what we can be the best in the world at (specifically within the foundation), and what drives our economic engine.

Finally, we realized that we must retain faith that we will prevail in the end, regardless of the difficulties, and at the same time confront the most brutal facts of our current reality, whatever they might be. In Collins' book this is known as the "Stockdale Paradox."

What does all this mean to all of us as members of this Foundation (Family)? We are all passionate about our sons, each one of us has the ability to excel in



*Kate McCurdy addresses BSF's #1 priority, that of accelerating the movement of BSF's Science and Medicine program, with detailed underlying steps to accomplish this goal.*

some area of the foundation, whether it be in helping Kate McCurdy in Science and Medicine, spreading awareness, supporting the other families out there, contributing more to the Listserv, or raising funds.

As an organization, we have reached our five year goal in three short years. We are moving in the right direction and doing it in the right manner. But to truly be successful, we need to work together as a family, without fear but with

one common goal and interest -- finding a cure for Barth syndrome.

We truly are saving many lives through education, advances in treatment and pursuit of a cure. By being honest with ourselves and contributing to our full

potential, we will accomplish our goals. We all need this organization and this organization needs all of us.



*Twenty-seven Barth family members gathered in Steinhatchee, FL to come together to work on BSF's newly revised programs. The end result you may ask? A more dedicated and unified team of soldiers forging ahead to accomplish BSF's mission.*



# Family Services: Sustenance through Fostered Empowerment

By Valerie "Shelley" Bowen, President, BSF



Parenting a child with Barth syndrome represents the most complex role of a lifetime.

One must perfect the art of organization where that quality may never have existed before. One

must learn a new scientific language called medicine (in numerous subspecialties). One must learn to navigate complex (and often unfamiliar) systems — medical, educational, insurance, etc. One must learn how to be an active advocate for their child (having enough knowledge to ask the hard questions, make the tough requests and think outside the box). One must learn how to balance the needs of all family members, even when there are still only twenty-four hours in one day. One is blessed by being the parent of a beautiful child who just happens to have Barth syndrome. One must NEVER give up, even when darkness seems to prevail.

At BSF, we are resolute to provide information in a timely manner to our families. Because the majority of our most active volunteers are "seasoned" families, we are empathetic to our members' needs. Erudition is acquired and expedited for all constituents of BSF through fostered empowerment.

Support often rouses a negative implied synonym of need. Need comes in all forms:

- Need for information
- Need to access experts in the various components of Barth syndrome
- Need for a caring voice at the other end of the telephone

- Need to better understand the standards of care for a child with Barth syndrome
- Need to contribute
- Need to make a difference

At BSF, we deliver! Our desire is to see families find "normal," despite being in an abnormal situation. We assist families to find the answers they seek. We provide a forum where families obtain results with a click of the send button. Additionally, we all benefit from the energy and accelerated progress that comes from collaboration between families and scientists. We keep our families informed about progress in research. We also offer families avenues through which they can directly help advance knowledge about Barth syndrome by contributing to research, either with information or blood and tissue samples or by participating in specific studies.

The acronym for our Fostered Empowerment program is Fe, the symbol for iron. This is a strong element, in many ways symbolic of our BSF families. Education about Barth syndrome leads to creative, thoughtful discourse. Education stifles isolation and creates a new generation of "ironworkers" for BSF.



*Shelley Bowen visits with Moira Masterson, BSF Grandparent Liaison, during her trip to the Child Neurology Society conference which was held in Ottawa, Canada in October of 2004.*

# BSF Listserv: A Vital Tool For All Registrants

By Shelia Mann, Family Services, BSF



Shelia and  
Ben at BSF's 2004  
conference

The BSF Listserv community continues to be one of the most valuable programs for Barth families. In 2004, BSF purchased a new Listserv program to better meet the needs of our growing community. This program has a wonderful feature that allows members to search the archived messages, a valuable resource file which will provide immediate access to topics

previously discussed amongst other Barth families. To access these archives, go to: <http://peach.ease.lsoft.com/archives/BARTHSYNDROME.html>. Press any of the links, which will open a page requesting a login. If you are a registered participant, enter your e-mail address that is used for the Listserv, and your password that you created when you originally logged in (or click "get a new Listserv password" first, and create a password to login with).

Currently, BSF maintains Listservs for the following groups: Families; Physicians/Scientists/Other Professionals; Siblings; Grandparents; and affected Barth Boys. All serve as forums to exchange vital information and provide immediate access to informed opinions from other registered participants.

In 2004, the Family Listserv served 137 interested members and featured 47 topic leaders, which included physicians, educators, parents, genetic counselors and scientists. In an effort to provide every family with the opportunity to participate in the discussions, we have posted topics on a two-week cycle. An expert or parent cyber guest, who has dealt with issues pertaining to the topic, facilitates each topic discussed. These discussions have proven to be a valuable source of information to our families.

A complete list of featured topics on the Family Listserv in 2004 including the following:

1. Neutropenia
2. Barth Genetics & How to create a Pedigree
3. General Inquires about Barth Syndrome
4. Awareness in the Community
5. BSF Family Support Programs
6. BSF Fundraising Programs
7. BSF Science & Medicine Programs
8. BSF Blood & Tissue Bank
9. Cardiology & Barth Syndrome
10. P/T & O/T for the Barth child
11. Metabolic Abnormalities in Barth Syndrome
12. Gastrointestinal Issues & the Barth Child
13. Psychological Issues with the Barth Child
14. International Efforts & BSF
15. Heart Transplants & Barth Syndrome
16. Dental concerns and the Barth Child
17. The Pediatrician & Day-to-Day Issues
18. Open Discussion "How can BSF Improve?"
19. BSF Registry & Medical Records
20. Automatic External Defibrillators (AED)
21. Sibling Support
22. Education
23. Tips of the Trade
24. Grandparent Support
25. Hobbies & Activities for the Child with Barth Syndrome

As BSF grows and continues to search for other affected families, the BSF Listserv program will continue to help educate and inform our newly joined members, as well as existing members. If you have a family member who is not on our Listserv program, I encourage you to ask them to join. Please e-mail me, Shelia Mann, at [smann@barthsyndrome.org](mailto:smann@barthsyndrome.org) and I will gladly register them. Also, I am currently planning Listserv topics for the year 2005, and if you have any new topics that you would like to discuss please let me know. Thank you and I hope you continue to benefit from the BSF Listserv Program!



# A Summary of The Barth Syndrome Trust's Progress in 2004

By Michaela Damin, Chairperson, Barth Syndrome Trust

The Barth Syndrome Trust now has 25 member families within the UK and Europe. That represents 11 new families in the past year. We are continuing to work alongside the Barth Syndrome Foundation and are actively involved in developing our program goals within the areas of Family Services and Awareness. From a family point of view, we are committed to aiding the flow of communication between all our members worldwide, knowing as we do that sharing experiences is essential if we are to play a role in advancing better understanding about this disorder. We have also been attending conferences, making presentations and networking with related groups in order to raise awareness.

Instead of running off a long list of individual things that we have been doing, I thought that it might be of interest to focus on just a few of the activities we have been involved within the last few months...



## BARTH CLINIC – BRISTOL, UK

On June 17, 2004, the first-ever “Barth Clinic Day” was spearheaded by Dr. Colin Steward who is well known to all of us in the “Barth Community”. We had 8 families (with 10 affected boys) who attended this clinic. Finally these families had a chance to meet face to face; we had a morning of informative lectures and our children were seen by expert geneticists, hematologists and cardiologists in the afternoon. Valuable data were collected and the doctors at Bristol Children’s Hospital remain actively

involved in the diagnosis and treatment of affected children throughout the UK. The next clinic is scheduled for January 2005.



## FAMILY MEETING IN ROTTERDAM

On October 20, 2004 our Dutch and Belgian families had a chance to meet up for the day in Rotterdam. This was a wonderful opportunity for affected boys, adults and their families to all meet in person. The next meeting is scheduled for July/Aug 2005. If you would like more information about our next meeting, please contact Joke van Loo, our European Rep. at [jvanloo@barthsyndrome.org](mailto:jvanloo@barthsyndrome.org).

## NEWS FROM OUR FRIENDS AT DISNEY, LONDON...

### Rome Marathon – March 28, 2004

We are so incredibly fortunate to have the ongoing support of many Disney employees, spearheaded by Isabelle Lemettre. They have aided us in so many ways this past year and have been instrumental in providing us with the necessary funds to continue our programs. These are the words of one of the runners and fundraisers, Pio Cardoza, about the experience of running a marathon and raising funds for all our families. I think his sentiments were shared by the entire group of runners!

“You’ll be glad to know that on Sunday I completed the Maratona della Citta di Roma in 5 hours and 30  
*(Continued on page 38)*

## Update .. Barth Syndrome Trust (Continued from page37)



minutes. It consisted of excitement, anticipation, pain, more pain, even more pain, joy, desolation, desperation and at the end of it all, I produced the best impersonation of a robot, such was the stiffness in my body. Nine thousand (9,000) insane runners, remarkably little support from the Italian public, but running through incredible scenery in the most beautiful city in Europe....

I want to say a big thank you to you all for sponsoring me. Your contribution undoubtedly helped me through the most physically gruelling experience of my life. After running over 13 miles of the race on the cobbled roads of Rome with my fellow Barth fundraisers, the soles of my feet felt like they had been injected with lead weights. I started walking and it was at this point that I seriously thought of quitting. I spotted a really nice café on Piazza Navona and could see myself tucking into some wholesome spaghetti and downing a bottle or two of vino. Then I thought about you a lot [my contributors and the BST Families]. How could I show my face at Disney again after all the money you had donated to Barth syndrome? So I kept going... At the 39k mark, a couple of "Power Walkers" from Illinois slipped past me. This was the final insult. I vowed that I would start running for 2 minutes and walk for 1 minute until the end of the race; my mission was to finish the race in front of these walkers, even if it killed me. You'll be glad to know my mission was accomplished!"

The Rome Marathon raised an incredible £4443 for Barth syndrome (\$8,421US).

### **GUY-FOX PARTY – November 6, 2004**

After the gruelling marathon, Isabelle decided that her next fundraiser would be a little less strenuous, so she decided to organize a party in Kensington, London with all the proceeds going to the Barth Syndrome Trust. Once again, Isabelle and her colleagues did everything to ensure that we had a great time whilst raising funds for our favourite cause. In fact, everyone had such a good time that they are all asking when she is planning her next party! Our good friends raised an amazing £2500 pounds (\$4,737US) from this evening.

A special note of thanks goes to Annick & Rob Manton and Terri Allison who head a formidable bunch of volunteers in the Overton area. Together they have organized parties, musical evenings and raised £773 (\$1,465US) for us. On December 11<sup>th</sup>, we will be arranging a Christmas Fayre in Overton, Hampshire which I am sure will be a great success!

And since this edition of the newsletter will focus on our International 2004 Conference, I would like to thank the Children's Heart Federation, Heart Transplant Families Together and the various Lions Clubs, Rotary Clubs and Round Tables. The donations from these organizations were of tremendous help to our UK families who attended the last conference. So these are some of the highlights of the past few months. We have been incredibly busy and enjoying every minute of it. If you live in the UK or Europe and would like to become more actively involved in any way, please contact me at [mdamin@barthsyndrome.org](mailto:mdamin@barthsyndrome.org).



### **BARTH SYNDROME TRUST (U.K. & EUROPE)**

Registered Charity Number 1100835  
(An Affiliate of The Barth Syndrome Foundation, Inc.)

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# UPDATE FROM BSF OF CANADA

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*By Barth Syndrome Foundation of Canada Board of Directors*

Hello and greetings from the Barth Syndrome Foundation of Canada (BSF Ca). Reflecting on the things to include in this update, it is amazing to see the progress that we and the rest of the "Barth syndrome family" have achieved. The conference in Orlando this past July was a tremendous success. New insights, such as the muscle tightness in some of the boys and young men, were noted. Research into the cardiac component through the ECG's, echo's, T-Wave Alternans and holter monitoring was furthered. And of course, the networking of families, the "What did you do when...?" or the "Have you ever experienced...?" was of tremendous importance. For those of you who were able to attend, it was a joy to reacquaint with old friends and a pleasure to make new ones.

With the conference over, it was time for the acting board to return to the business at home. Over the summer, a preliminary draft of our charity application was done as well as a search to find those who would be responsible for the auditing of our books. We are pleased to announce that Madgett, Roberts, Marlowe, Jackson & Associates have been appointed our auditors. We now have audited financial statements for 2003, and our charity application has been submitted. We hope to receive our charity status early in the New Year.

We have also been working on plans for our first Annual General Meeting to take place sometime in February or March, which will be held by teleconference. During this meeting, official elections of the board will take place as well as presentations of financial reports and program updates. Details will be mailed to those members who are asked to take part. Concurrently, Lynn Elwood, has been working on the Canadian website [www.barthsyndrome.ca](http://www.barthsyndrome.ca) which is now up and running.

BSF Ca participated in supporting the Wisconsin Ironman this past September. John, Gary and Tim did a tremendous job, and our heartfelt thanks and

appreciation go out to them. Their sacrifice and commitment on behalf of our sons is incredible. The triathlon was a success, and we are thankful that we were able to contribute. Thank you to all the Canadians who made pledges. Also on the fundraising front, Cathy Ritter is once again leading us to record sales in our holiday poinsettia sale, and plans have begun for the First Annual Canadian Barth Syndrome Charity Golf Tournament to be held September 2005.

We have undertaken an aggressive awareness campaign over the last few months. Cathy Ritter, Lynn Elwood and Shelley Bowen ran a booth at the Child Neurology conference in Ottawa in mid October. The conference went well, and a considerable number of neurologists were added to BSF's mailing list. With the help of Lois Galbraith, we have done a phone and mail campaign contacting Children's hospitals across the country. We now have resource centres in Children's hospitals across the country receiving this newsletter and a number of specialists in a variety of hospitals. This campaign will continue in the New Year to raise awareness and to endeavor to find new families to join BSF Ca.

Family support continues with telephone conversations to non-internet based families as well supporting those families who have contacted us with a possible yet unconfirmed diagnosis of Barth syndrome. To date, most of these families have not ended up with a Barth diagnosis.

We will continue to work on our core programs of awareness, family support, science and medicine and the fundraising used to help support these programs. We were active participants at BSF's international planning session held in November and are participating in a number of international initiatives. There is a great deal of work to do to help our boys and young men. Please get involved in any way, small or large. Let us know your thoughts and ideas. We look forward to hearing from you!



# The Canadian Summer Gathering

By Lynn Elwood (Ajax, Ontario, Canada)



*The Canadian families gather outside of Lindsay for a day of fun, otherwise described as a day "good for the soul"*



*Travis, Jared, Adam and Robert enjoy a day together with other Barth families located in Canada*

The August day was sunny and cool when some of the Canadians made the trek with a van full of excited kids, food and family to Lindsay, about an hour to the east of Toronto. The Hones had come to town from Saskatchewan and set up a gathering for the nearby Canadian families. The Gordon (with Grandmother Moira), Hope and Elwood families were able to make the trip.

Thanks to the hospitality of Chris' parents who live on a working farm outside of Lindsay, the day could not have been better. There was a horse and cow to see, lots of kittens, a huge garden and most fun of all, a giant hay loft. When we arrived most of the kids quickly disappeared into the barn where they jumped and played in the hay until they were called out.

There was a pool for the daring kids to have a quick and cool swim in, and an all terrain vehicle to take turns driving (or flipping in Robert's case). Jared and Andrew sat comfortably under the awning and shared the fun. Later there were tunes on Travis' guitar and a fire to warm our hands by. As with every other Barth related gathering I've been at, the kids got along famously, behaving perfectly and letting the parents enjoy a good talk.

We had a truly wonderful day, the kind of time that we call "good for the soul". It was great getting to catch up with Susan, Chris and the Hones and to have all of our families and kids together. I have to thank our Barth boys for once again bringing us together with a special group of people, and of course the Hones who arranged it and made us all feel so very welcome.



## *Canada sells Poinsettias!*

Once again this year, Cathy Ritter has organized a poinsettia sale to raise funds for BSF of Canada. The orders have been taken and we're awaiting delivery of almost twice as many plants as we sold last year. This has proven to be a very successful fundraising project with an expected profit of over \$2,000. We've raised awareness and much needed funds, and we're having fun at the same time. Thanks to everyone that has been selling and organizing. Special thanks to Cathy Ritter, Karen Gordon, Audrey Hintze and Heather Reppen, our top sales people.

# Finding One's Special Niche

By Lois Galbraith (Ontario, Canada)



*Lois Galbraith (right), partners with Maria Olson, while manning the registration booth at BSF's 2004 conference.*

Attending the 2004 Barth Syndrome Foundation's Conference this past July was enriching and emotional for me, and all of this was possible because of the birth of a special child just fifteen years ago this December.

Adam came into our lives and Les and I had our very beings changed. When he was diagnosed with Barth syndrome, he was still very special and a gift. We would learn so much more about ourselves and the world through his eyes. We began to help by "baby-sitting" Adam for periods of time (NO hardship there), and we remain grateful that Lynn and Rick are wonderful about sharing him.

I began hearing about the American Barth group and a "Shelley Bowen". I read the "Saving Michael Bowen" article and soon began listening-in on the Listserv. The eloquent words of Shelley began to stir my "need-to-help" genes. I was compelled to write her a letter.

Cathy, Chris, Karen and Lynn began the formation of the Barth Syndrome Foundation of Canada, and I was awed by the strength, dedication, time and love that was being poured into this. There was talk of a third conference... I had to do something.... "If I can help with anything just let me know!!"

Soon I was on the internet contacting doctors about their bios/photo/consents/etc to be included in the conference program. I met the very lovely Kate McCurdy (by phone first). She was so patient,

helpful, encouraging and thankful. She was helping Will to deal with his problems at the time and still had time for me and my persistent questioning. "Shelley would like some help with clinics - Anna could use some assistance on the site...". How wonderful and buoyed I felt to be needed.

I went to Orlando and the Coronado and watched the group behind the registration desk for ten minutes or so. Then it was ..."Hi, my name is Lois Galbraith and I'm..." when Sue jumped from her chair, extended her hand and said "I'm Sue Wilkins, welcome...so great to meet you!" The ice was broken and the fun began.

I am an observer of life, and the next six days were like a field-day!! Meeting all of the Barth boys/men was the most compelling and awesome experience for me. I eyed them, I spied them, I loved them!! I was equally overwhelmed by the dedication of the physicians, the clinicians, the educators and all of the volunteers. I watched parents hug each other and the boys, I watched the boys/men interact so freely. I watched the Conference committee attend to every detail with love and caring. I watched the group come together for social times, for photographs, for meals and for the culmination. These were touching, emotional, "gasp-for-air" times for me.

"So what is it about BSF that has captured your heart and soul?" Lynda asked. It is all the extraordinary people beginning with Adam, my supportive husband Les, and of course the loving Lynn. Just to seal the commitment you find Shelley, Kate, Anna, Sue, the doctors, educators and volunteers and you see these people coming together for our esteemed Barth boys/men! Wow - blows me away!! I have been so very enriched by everyone from Adam to the Barth syndrome world of families. God Bless!



# BSF Update From South Africa

By Jeannette Thorpe, South African Ambassador, BSF



Jeannette Thorpe (right) and Michaela Damin (left), are two of BSF's international leaders

Well, this has been a fairly eventful year from South Africa with a couple of exciting happenings. An awareness campaign targeted physicians and professors in the areas of: immunology, hematology, genetics, endocrinology, pediatric cardiology, pediatric neurology

and various other heads of departments. Many of the above-mentioned doctors were secretaries or chairpersons of their area of specialty, and I appealed to them to forward the information provided onto any physicians registered with them or any other individuals that may have an interest in Barth syndrome.

To continue in my efforts of raising awareness of Barth syndrome, I received permission to display BSF's newsletter at the KIDZ N' ALL Conference, a large pediatric conference held in Cape Town during August of 2004, as well as the South African Heart Congress, a large conference covering a broad range of cardiac interests, including pediatric cardiology, which was held in Durban in October 2004. BSF Scientific brochures were also placed into congress bags.

I gave a presentation to two pediatric physical therapists on Barth syndrome and hope to be able to present at one of their meetings to a larger group.

**A major accomplishment has been met**, and we are now able to conduct genetic testing for Barth syndrome in South Africa. This comes with great thanks and gratitude to Dr. Fraser Pirie and Molecular Biologist Professor Rosemary Pegoraro – both of whom were responsible for coordinating and setting up this facility. A huge thank you also

goes to Dr. Iris Gonzalez who gave us much assistance in getting the testing off the ground! The tests are conducted at the Nelson R. Mandela School of Medicine, University of KwaZulu, Natal. Barth syndrome screening will be placed on the National Genetic Screening database for the whole of South Africa to use.

Nigel, Thomas, Benjamin and myself attended the 2004 Barth Syndrome International Scientific/Medical and Family Conference at Disney's Coronado Springs Resort. This was a humungous WOW on all fronts! Everybody's needs were catered to. Individual agendas were organized for the scientific and medical attendees, parents, siblings (who were divided into age appropriate groups) and Barth boys (who were also divided into age appropriate groups). Not only were there educational programs organized, but also some wonderful social events. Well done to everyone involved in putting this incredibly inspiring event together. I would like to also take this opportunity of thanking all those involved with the research of Barth syndrome – you are truly a lifeline to our group! *I highly encourage families to attend our next conference – you won't be disappointed!!*

We continue to work toward obtaining our fundraising number and trust set up before the end of the year.

Family support remains a pretty quiet area for us here in sunny South Africa as there are still only two known cases (ie., Benjamin and my sister's son Colin). I continue with complete dedication to this incredible group and most especially to our Barth boys! Should anyone like to comment, volunteer or need further detail, please contact me at: [jthorpe@barthsyndrome.org](mailto:jthorpe@barthsyndrome.org).

We have a South African web page – if you would like to have a look, please go to: [www.barthsyndrome.org/South\\_Africa.html](http://www.barthsyndrome.org/South_Africa.html)



# OUTREACH PROGRAM CONNECTS FAMILY TO THE BARTH SYNDROME FOUNDATION

By Michelle (Texas)



*Michael Anthony (son) with parents Michelle (Mom) and Michael (Dad)*

Our son Michael Anthony Jr. was born on November 25, 2001. Although Michael was full term, he only weighed 5 lbs. 9 oz. We took Michael home the following Tuesday, but returned to the hospital Wednesday morning because he would not eat and became dehydrated. After a short

stay, Michael got better and we returned home. Everything seemed fine for the next year.

On March 31, 2003, Michael became very lethargic and he fell asleep while sitting in his high chair. We ended up in the ER at Brackenridge Children's Hospital. Several doctors examined Michael but could not determine what was wrong with him. They could only tell us his heart was enlarged and he was very sick. We were in the PICU for several more days and saw a multitude of specialists. Michael was tested for every possible type of virus, bacterial infection, and disease. One doctor mentioned the possibility of Barth syndrome, but the others felt that this would be highly unlikely because it was so rare and we had no family history. They tested Michael anyway. After a few days, we were informed that Michael had tested positive for Barth.

Determined to find out anything and everything we could about Barth syndrome, we purchased a laptop computer so that we could access the Internet from the hospital. We contacted the Foundation to notify them about our positive diagnosis. The following day, we received a telephone call in our PICU room! The information and support they gave us was tremendously helpful. On Sunday, April 20, 2003, Easter Sunday, Michael was released from the hospital, with a few medications and a positive diagnosis.

Over the next year, we became members of the Barth Syndrome Foundation. We were also added to the Listserv, which is our lifeline to the other families and doctors. In the beginning, we just tried to absorb all of the information. Attending the Texas Outreach Meeting in Brenham, Texas and then the Conference in Florida, has had a life-changing impact on our lives.

At BSF's July 2004 conference, we were finally able to meet all of the families that were making the posts on the Listserv. It was comforting to actually meet parents who had gone through the same emotional roller coaster that we had. The conference also gave us the opportunity to meet other boys and young men with the same illness as our son. They are truly an inspiration and have touched our hearts forever!

We discovered a group of people who did not even know our son, but were giving everything they had to help him. They share their time, money, support, and complete dedication to give him and every boy with Barth syndrome a better life and they are working to find a cure. We thought these people were amazing and we wanted to be a part of them.

Michael and I are dedicated to helping BSF in any way we can. If we are asked, the answer will always be yes. We have volunteered to be mentors to new families, help with the planning of BSF's 2006 conference, and we have joined the family support and awareness committees. We are committed to telling anyone who will listen about Barth syndrome.

We were fortunate that our son was diagnosed early. Our doctor's awareness of Barth syndrome helped our son, and we want to do what ever it takes to help others become aware. We want to support families that have been diagnosed and to find those that have not yet received the correct diagnosis. We feel we all have something to contribute, and we can all make a difference!



# BSF Conference Helps Family Prepare

By Greg (Texas)



Top row: Kristi (12), Keli (Mom), Kati (12)  
Middle row: Jessi (10), Shayli (4), Shelbi (6),  
Bottom row: Joshua (9) and Greg (Dad)

**K**eli and I always wanted a big family. We were high school sweet-hearts and, even then, spoke of wanting at least six children! That desire has never wavered.

As childbearing began, the Lord blessed us with twins and we never looked back. During a period of eight years, we received into our family six healthy children. Having babies seemed to go like clockwork. We never dreamed of a difficulty.

That naivety all changed with baby number seven. At birth, Caleb presented with a number of difficulties and was flown to Texas Children's Hospital in Houston. He spent two weeks there, but came home without a diagnosis. The doctors could only say that he seemed to be "better".

Though he had a variety of symptoms during his short life, no one was able to diagnose the root cause. His medical care always involved a lot of head scratching on the part of the doctors. At about fourteen months of age, it was discovered that Caleb's heart had enlarged to the point of no return. He was immediately flown to Lubbock, where he struggled for three days before going to be with the Lord. It was, to this point, the most difficult day of our lives.

Keli relentlessly set out to discover what the doctors had been unable to learn. She spent countless hours researching his condition. As a result of her persistence, she came to believe that he probably had Barth syndrome. Genetic testing of his tissue confirmed her belief.

We were at once relieved (to finally have a diagnosis), saddened (at not having known it sooner), and frightened (because we were, by then, expecting another child). We didn't yet know the sex of our unborn child, much less whether or not the mutation was present. We arranged for an amnio and then set out to learn what we could about Barth.

In July, we attended the BSF conference in Florida. At the time of the conference, we had not received the test results. We just knew that we needed to be there and learn all that we could. After returning home, we found out that Benjamin did indeed have the mutation.



Benjamin was  
born on October 6, 2004

Benjamin surprised us by arriving five weeks early, but presented well. His heart function was a bit low, but he was immediately started on the heart medications and responded well. He continues to grow and thrive, cared for by a

loving family and a group of well-prepared doctors.

We're so thankful for having attended the BSF conference! It gave us the information we needed and also allowed us to meet a wonderful, loving group of families. We look forward to participating in BSF so that others will benefit as we have!



# BSF Sibling Spotlight

By Jess Wiederspan (Lincoln, Nebraska)

This new section of the newsletter will feature our awesome BSF siblings. Please e-mail any comments or suggestions to Jess Wiederspan at [jesswieds@yahoo.com](mailto:jesswieds@yahoo.com)



Mary Kate Baffa shows off her hula-hooping skills

## Mary Kate

**Full Name...** Mary Kathrine

**Sibling of...** Kevin (15)

**Location...** PA

**Age...** 10

**Grade in school...** 5th

**When I grow up, I want to**

**be...** a chef, a makeup artist, or an interior designer

**My favorite subject in school is...** recess

**If I could travel to one place I've never been, it**

**would be...** Europe

**In my free time, I like to...** play on the computer

**One thing I am really good at is...** sleeping

**My favorite food is...** Chocolate

**My favorite book is...** *Because of Winnidixie*



Thomas Thorpe hangs out with Dale, his brother Ben, and Nicholas Damin.

## Thomas

**Full Name...** Thomas

**Sibling of...** Ben (11)

**Location...** South Africa

**Nickname...** Thorpy, Doup, Thorpedo, T-Biscuit

**Age...** 12

**Grade in School...** 6th

**When I grow up, I want to be...** an actor

**One unique thing about me is...** I am really impatient but I still play golf

**If I could travel to one**

**place I've never been, it**

**would be...** Australia

**In my free time, I like to...** play golf, play the drums, and chill out

**One thing I am really good at is...** imitating people

**My favorite food is...** Alfredo



Olivia and Kelsey determine how many licks it takes to get to the center of their Mickey pop !!

## Kelsey

**Full Name...** Kelsey Elizabeth

**Sibling of...** Jack (12)

**Location...** NJ

**Age...** 13

**Grade in school...** 8th

**When I grow up, I want to be...** an interior decorator

**My favorite subject in school is...** gym

**If I could travel to one place I've never been,**

**it**

**would be...** Australia

**In my free time, I like to...** talk on the phone and hang out with my friends

**One thing I am really good at is...** field hockey and making people laugh

**My favorite food is...** Nachos

**My favorite book is...** *Three Girls in the City* series

## **Quotes from Sibs**

### **Who Attended BSF's 2004 Conference**

"It was incredible for me to be in the same room for the first time with a group of people who all had a brother with Barth syndrome. For once, I didn't have to try and explain what Barth syndrome was. The conference was an amazing and eye-opening experience for me and I am excited to become more involved in BSF in the future." ~ Jess, age 25

"I liked the marshmallow meeting (where we learned about the genetics of Barth syndrome) and the Sib Shop. Donald (Sib Shop Leader) was cool!" ~ Mary Kate, age 10

"I was really amazed by the wisdom and maturity that the siblings and affected boys displayed at the conference. I really felt that I learned a lot from them; with these kids as the future of BSF, I have no doubt that the organization is going to be in great hands!" ~ Alanna, age 25

# The World of Ferrets

By Adam, Age 14



*Adam cuddles with his ferret friends, Wilma and Betty!*

When I come home from school I like to take my ferrets out and cuddle with them. They climb up and curl round my neck. They play with each other and with me. Most of all they cuddle with me.

When ferrets play they bounce up and down and bite each other. If you didn't know them you'd almost think they were hurting each other but they are not. They have very thick skin.

Ferrets like to eat banana chips, raisins and dried papaya. Their regular food consists of basically chicken, corn, fish and some vitamins as well as a few preservatives. They require water every 15 minutes and food every 3 to 4 hours. They go to the washroom frequently.

Ferrets can be litter trained but it does not always hold, especially during car travel. They become disoriented and nervous. When they run around out of their cage, they tend to forget about the litter box and go wherever they find it convenient, such as corners.

I make sure their hammock is clean and clean the cage once a week. Every 1 to 2 months I bathe them. If ferrets are bathed more than once a month, it causes the smell to become greater. You must be careful around ferrets when sick because they can catch the flu and cold and they can give it back to you.

When you have ferrets, they help you control your temper because they sense your emotion and they will react with the same emotion. If you are angry they will bite and show anger. If you are happy they will show you affection and play with you. If you are sad they will become cuddly and sleep a lot. Overall I think ferrets are very good pets. While they are playful and inquisitive, they are also sensitive to how people react.



# Memories Cherished

By Robert, Age 18



*Robert gives a summary presentation at BSF's 2004 conference*

In July 2004, many of the Barth boys and their families from around the planet came, saw, and conquered Disney's Coronado Springs Resort. There was so much to do this time round that it was really impossible for anyone to get bored. The clinics

ran as a well-oiled machine, and the activities were well thought out and enjoyable. The location could not be improved upon, and for the most part the weather cooperated.

Some of the things that truly stand out in my mind include the massive pool, the incredibly hot weather mixed in with thunderstorms, the Mission Space ride at Epcot, and of course, the dreaded Microvolt T-Wave Alternans Test (a/k/a torture via sandpaper).

The best times that I can remember are being able to get together with what are now good friends, talking to them again, going for a swim and to the Downtown Disney Quest arcade. The "hang time" was most memorable.

At the Saturday night social/luau, what could be more amusing than some of the guys putting on grass skirts, watching our respected doctors doing the limbo, and our mothers recapturing their youth trying to do the hula hoop. And who can forget Michael's Goofy hat?

Jon Rosenshine's moving slide show presentation on his laptop brought the conference to a close back on the floor where we started. Lingering memories include dissecting split peas to learn about DNA, and making a presentation to everyone with some advice for the younger boys. I left the conference more informed about Barth syndrome, and about what the future can hold for us truly unique individuals.



# Barth Boys

## A Poem Written by Daryl , Age 23

Barth Boys, we have learned so much,  
and now it's time  
to take what we have learned and use it.

Let people know what you know.  
because no one knows more about your body than you.  
We showed the doctors a thing or two,  
using words and knowing things only doctors should know.

Barth Boys are different and we all know that now.  
But the difference is that we are special.  
People may not understand the reason why but God made us this way.  
But don't let people tell you what you can't do;  
If we don't try we'll never know if we could.

Put your mind to it even if you don't think you can...TRY.  
You know if you can do it or not.  
We all know our own limitations and how far we can push ourselves.  
But as long as we try that's okay...  
At least you tried and no one can judge you for that!

Us Barth Boys have a promising future.  
With ICD's and all the other research, we will survive.  
And with the loving help and support in a timely manner,  
we can make The Barth Syndrome Foundation known worldwide!

### WORDS TO FIND

BARTH  
BTHS  
CARDIOLIPIN  
CARDIOLOGY  
CARDIOMYOPATHY  
CARRIER  
DELETION  
DNA  
GENE  
GENERAL FATIGUE  
GENETIC  
GROWTH DELAY  
HEMATOLOGY  
MEMBRANES  
METABOLISM  
MUSCLE WEAKNESS  
MUTATION  
NEUROLOGY  
NEUTROPENIA  
NEUTROPHIL  
SYNDROME

## Barth Syndrome

L	I	H	P	O	R	T	U	E	N	M	D	K	A	E
T	A	F	F	A	Z	I	N	W	D	U	E	N	O	M
R	E	I	R	R	A	C	Z	I	F	S	D	T	T	O
G	E	N	E	T	I	C	D	R	A	C	G	G	Z	S
E	N	E	G	C	A	R	D	I	O	L	O	G	Y	O
E	U	G	I	T	A	F	L	A	R	E	N	E	G	M
L	C	K	Y	A	L	E	D	H	T	W	O	R	G	O
N	E	U	R	O	L	O	G	Y	L	E	I	L	S	R
C	A	R	D	I	O	M	Y	O	P	A	T	H	Y	H
M	E	T	A	B	O	L	I	S	M	K	A	B	N	C
C	A	R	D	I	O	L	I	P	I	N	T	A	D	X
Y	G	O	L	O	T	A	M	E	H	E	U	R	R	S
S	E	N	A	R	B	M	E	M	H	S	M	T	O	H

# In Loving Memory

## Caleb Michael Holly

June 4, 2002 - August 8, 2003



***Behold, children are a gift  
of the LORD; the fruit  
of the womb is a reward.***

*(Psalm 127:3)*

It is an unnatural thing for a parent to outlive a child. It is, perhaps, the greatest injustice of all. And yet, we know that every child is a gift of the Lord and precious in His sight. Within each gift we find blessings.

Our Caleb spent only fourteen months with us, yet touched us for a lifetime. He was the baby brother to six loving siblings. They adored him. We all did. His blue eyes, long eyelashes and full cheeks were crowd pleasers. He attracted attention everywhere he went.

Caleb loved music, animals, the outdoors, and rolling his ball. But, most of all, he loved his Mommy. Most of his time was spent in contact with her. His feet rarely touched the ground.

It's been a year and a half since we lost Caleb, and now we have a new Barth boy (Benjamin). Benjamin fills our arms and our hearts in a way that comforts us, yet without replacing the memory of his sweet, older brother. God is so good.

We still grieve for our loss, yet celebrate the victory of everlasting life. Like the great King David of old, who also suffered the loss of a son, we determined to wash ourselves, take food and worship the Lord. We cannot bring him back, but we will go to him. (see 2 Samuel 12:16-23).

*In loving memory of Caleb,  
His Daddy, Greg Holly*



## Gabriel Antomarchi

February 19, 2004 - September 4, 2004

"We would like to thank all the members of the Barth Syndrome Foundation who helped us during Gabriel's disease and after his loss.

Through this foundation we found some hope and the strength to face this disease, allowing us to enjoy our curious and cheerful baby."



Best regards,  
Eva and Nicolas  
Antomarchi

Nicolas travelled many miles to attend BSF's 2004 conference, while his wife Eva and son Gabriel remained at home due to Gabriel's unstable condition. To avoid a language barrier, an interpreter was provided to Nicolas throughout BSF's 2004 conference via the Alliance Francaisae de Orlando group.

On behalf of BSF, we offer those who have lost children our sincerest condolences, and we pledge our commitment to remain a strong support to these families.

At BSF, we take great pride in the services we provide to all our families, and despite the fact that we are aware of the loss of two children to Barth syndrome in 2004, we are more adamant than ever before to reach our Vision, "...A world in which no one will suffer or perish from Barth syndrome."



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*Seven year old Jacob, son of Darlene and Alex of Texas,  
smiles jubilantly while embracing the spirit of giving!!*

## **VISION**

***Today, Barth syndrome is a rarely understood, frequently fatal, genetic disorder affecting boys.***

***The Barth Syndrome Foundation's Vision is...***



## **MISSION**

***The Barth Syndrome Foundation's Mission is ...***

***"To guide the search for a cure, to educate and support physicians, and to foster an informed and caring community for affected families".***

***"A world in which no one will suffer or perish from Barth syndrome"***

The Barth Syndrome Foundation, Inc. is the only group in existence globally in which families, physicians and scientists work together to address Barth syndrome – a rare but serious X-linked recessive disorder comprised of: cardiomyopathy, neutropenia, skeletal muscle weakness, exercise intolerance, extreme fatigue and growth delay, in varying degrees. Since early diagnosis is key to survival of Barth boys, BSF strives to save lives through education, advances in treatment and pursuit of a cure. The organization offers information to physicians and families, promotes awareness, encourages and funds research, maintains an international registry of Barth patients, and provides a caring support network for all those affected by this condition.

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



**The Barth Syndrome  
Foundation, Inc.  
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