



SPECIAL COVERAGE OF BSF's 2002 CONFERENCE

A LETTER FROM 'BSF's' PRESIDENT

Dear Fellow BSF Members,

This publication cites the many accomplishments made during our most recent conference. The BSF Family/Scientific Conference is clearly among our greatest tools to deliver a better understanding about Barth syndrome to families and scientists alike. In this edition you will read articles from scientists, parents, siblings and grandparents. This was an event that we can consider a successful springboard to move forward.



The BSF conference is consistent with supporting our mission of *guiding the search for a cure, educating and supporting physicians and creating a caring community for affected families.*

While BSF has made a difference in providing life-saving measures to improve the mortality rate of children affected by Barth syndrome I must also add my overwhelming concern over the loss of four children during the course of the past twelve month cycle. This should serve as a stark reminder that we are still dealing with a far too frequently fatal disorder.

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BARTH SYNDROME ... A MUCH MORE COMMON DIAGNOSIS THAN GENERALLY RECOGNIZED

By Colin G. Steward, Ph.D.

“By now, I was beginning to suspect that this disease was not as rare as was generally thought.”

Dr. Colin Steward is Reader in Stem Cell Transplantation at the University of Bristol and a Consultant in Bone Marrow Transplantation for Children with Metabolic and Genetic Diseases at the Royal Hospital for Children in Bristol. Several years ago he and his colleagues published a paper in the *Journal of Pediatrics* explaining that they thought Barth syndrome to be a much more common diagnosis than was generally recognised at the time, perhaps because affected children could present to many different medical specialists. They have since gone on to identify other children from the South of England, confirming their belief. We asked Dr. Steward to speak on this topic at the conference. He presented his findings as a narrative in the hope that families and doctors could get a feel for ways in which the diagnosis can be missed and the potentially tragic consequences. For those of you who could not hear it, here is The Bristol Story in Dr. Steward's words.

A Chance Encounter

I first became aware of Barth syndrome shortly after I began work as a Consultant working with children with genetic diseases. A colleague, Dr. Steve Lewis, bleeped me to ask advice about Brandon, a 15 month-old boy whom he had just seen in the outpatient department. He explained how Brandon had been transferred to the Intensive Therapy Unit (ITU) several weeks before, having been put on a respirator because of a severe attack of croup

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LETTER FROM 'BSF'S' PRESIDENT

(Continued from Cover Story)



Michael Bowen visits Dr. Barth's homeland

BSF's vision is a world in which not one more child will suffer or perish from this condition. The loss of these children substantiates the need to remain focused on our ultimate vision while supporting our mission statement.

I surround myself with photos of all of your children. While I work on BSF issues I need only glance over from my computer monitor to see a photograph of these children to become even more impassioned about our work. I also have a picture of my children on my desk to remind me that this work is initially

driven by the loss of what could or even should have been for our darling son Evan and the drive to ensure a better future for our beloved Michael. We should be appalled, grief-stricken, angry and pro-active in asserting ourselves to make sure this disorder not claim one more life!

Having known the devastation over the loss of a child, I remain determined to ensure a legacy is made for each of the children we have lost to this disorder. The impact they have made in their far too brief lives extends beyond those who knew and loved them, but also onto future generations of children and families who will be affected by this disorder. Many of our parents have channeled the agony over the loss of their child into a passion for sparing yet another family from enduring the same loss. I commend them for their overwhelming bravery to overcome the pain and for their selfless effort to immerse themselves in working with BSF to help other families. We, as an organization, can make a difference in the future of this disorder. I charge our families to become more involved in various ways. Most importantly, take the time to participate in BSF's registry, which serves as an exceptional, perspicuous tool of the various components of this disorder. This is a rudimentary yet most important step to provide much-needed insight to scientists about how Barth syndrome manifests itself in our children. We have all been grief-stricken over hearing of the loss of these children. The greatest honor we can provide them and their families is to provide our input through BSF's registry.

I also charge our scientists and clinicians to network and assist us in identifying ways to better care for these children. Answer questions that are posted on the listserv and help us to create guidelines in the management of care for these children. Assist other physicians in delivering the best possible care for these children. Assist us in our awareness program to promote knowledge about Barth syndrome to insure a timely diagnosis. Continue to collaborate with each other, and help us to reach our ultimate vision.

Sincerely,
Shelley Bowen

Shelley Bowen
President, The Barth Syndrome Foundation, Inc.

BARTH SYNDROME... A MUCH MORE COMMON DIAGNOSIS THAN GENERALLY RECOGNIZED

(Continued from Cover Story)

at his local hospital. His doctors became concerned because he seemed excessively floppy and did not seem to be improving at the expected rate. They began to think that Brandon might have a serious muscular disorder, to explain why he had coped badly with the virus, and asked our ITU to take over his care. However, by the time he arrived in Bristol he seemed to be much improved and actually came off the respirator on the following day.

The main thing which concerned the doctors in Bristol was that his blood count was abnormal. The machine in our hospital routinely splits up the white cell count into its respective components (neutrophils, lymphocytes, monocytes, etc.). This showed that he had a severe lack of neutrophils, his count being 0.1 compared with a normal of 1.5 or above for a white male infant of his age. The ITU team asked the senior haematology consultant to review Brandon and a bone marrow was performed. This looked slightly abnormal, but not sufficiently so as to suggest a serious diagnosis and the changes were thought to result from the viral infection. Soon after this he was discharged and went home completely well with an appointment for review several weeks later, which is where my colleague came across him. His neutrophil count was still extremely low but Steve was also concerned to hear about Brandon's only brother, who was sick from birth with fits, feeding problems and slow development. He was hospitalised for many weeks after his birth, but eventually was able to go home on nasogastric tube feeding. At six months of age - when he finally seemed to be putting on weight and starting to develop - he was found dead in his cot without any warning. The cause for his death was never clear, not even at post mortem examination.

Steve asked if I could think of any diseases which might run in the male line of family, cause muscular weakness, severe neutropenia and even result in death. My only thought was that this might be due to a mitochondrial disease (mitochondria are the powerhouses of cells and are very important in muscle function) and I suggested that he should ask the opinion of the hospital's Consultant Chemical Pathologist, Dr Charles Pennock. On hearing the story Dr. Pennock immediately said "It sounds like Barth syndrome" and went on to describe what he meant.

Dr. Pennock explained that it was an X-linked genetic disease (passed on by the women in a family but only causing disease in the males) which caused low neutrophil counts (neutropenia), abnormal heart muscle (with a thickened inner lining, termed endocardial fibroelastosis), weakness of arm and leg muscles and excessive excretion in the urine of a chemical in called 3-methylglutaconic acid (3-MGC). Steve was amazed: he had just been reading a letter about the little brother who had died in his cot. The baby had undergone extensive investigations for metabolic diseases whilst he was very young but only one of the tests had ever been abnormal - an excess of 3-MGC in the urine. His doctors had carefully investigated this problem, giving him a "leucine loading test" to see whether this caused further elevation of the abnormal acid excretion. It did not, but neither was the 3-MGC level raised on a subsequent test. A differential white blood cell count had not been performed so that we cannot know whether he had low neutrophils.

Children with Heart & Neutrophil Problems

As a doctor regularly asked to see children with low neutrophil counts, this concerned me. Here was a disease which could cause severe abnormalities and - from

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Back issues are also available.

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what I later read in Professor Barth's paper - carried a real risk of infant death from either heart failure or sepsis, yet I had never even heard of it. However, I was heartened to see that the disease was stunningly rare, having been described in just one Dutch family and a very few other cases throughout the entire world. By chance I met Dr. Pennock shortly afterwards and asked him where he had come across Barth syndrome. He explained that one of the cardiology consultants had identified a child with the disease and went on to tell me about Matthew.

Matthew did not thrive during his early months of life and then had developed severe heart failure at six months of age. He was started on medicines such as diuretics (to reduce the amount of fluid in his circulation and hence reduce the strain on his heart) and digoxin (to strengthen the pumping of his heart). Initially he responded well; however, in the middle of his second year he deteriorated so badly that he needed a heart transplant. The heart which was removed showed severe endocardial fibroelastosis, but no diagnosis was suggested. He had a torrid time in the early months after his transplant with multiple infections including several episodes of severe chickenpox. During this he developed a low neutrophil count. The haematology team were consulted, a bone marrow was performed, which showed minimal changes and it was all assumed to be a response to his severe viral infections. Eventually he recovered and was discharged from hospital. It was only several years later that his cardiology consultant came upon Barth syndrome in a medical journal and made the connection. Further investigation showed that his neutrophil count was often low and that he had an excess of 3-MGC in his urine.

Out of curiosity I started to ask around. Another of the other cardiology consultants did not know of Barth syndrome, but did have a patient - Anthony - on whom she had sought a haematological opinion. He had a truly remarkable history. At 2 months of age he had undergone surgical repair, under general anaesthetic, of a small hernia in his groin (an "inguinal hernia"). This went without incident, but just 9 days later he presented in severe cardiac failure and was found to have a neutrophil count on various tests ranging from 0.2 to 0.7. An ultrasound examination of his heart (echocardiogram) showed a swollen heart (dilated cardiomyopathy). His neutropenia was investigated but no cause was found. It was thought that his low count was simply due to depression of the bone marrow by the same virus which had damaged his heart (although the virus concerned was never identified). He too was treated with drug therapy and had a

very good response. We subsequently found that he had an excess of 3-MGC in his urine as well as recurrent neutropenia.

Just after we finished investigating Anthony a 16-month-old boy was brought into the ITU with severe heart failure and X-ray signs of a chest infection. This boy's first measured neutrophil count was 0.6, although many subsequent counts were in the normal range. His parents, who were both from a medical background, were able to confirm that his health had been unremarkable apart from being small for his age and seeming rather weak in recent months. Again we wondered whether a virus had damaged his heart, especially since the low count was only transient, but a urine test showed that he also had excessive excretion of 3-MGC. Very sadly his heart failure could not be controlled and he died before he could receive a heart transplant. A post-mortem examination showed endocardial fibroelastosis, confirming the diagnosis of Barth syndrome.

Lessons from the Past

This led me to discuss post mortems with Dr. Michael Ashworth, a Consultant Pathologist. I asked whether there might be previous boys on record who had died with endocardial fibroelastosis. He explained that this seemed likely since the department's records went back for decades. So he looked back and found one suspicious report on a boy called David, who had died at the age of 6 months in our hospital in 1959. His paediatric consultant, Dr Beryl Corner, had explained to David's mother that his post mortem showed his heart to be "shrivelled up, like a little old man's", but the cause of this had remained a mystery. This was a tragic family. In the same generation two other boys had died: one was stillborn and the other died of unexplained heart disease at six months of age. David's grandmother gave a heartrending story of how three of her four brothers had died as infants, all having been well initially but then turning blue due to sick hearts. [The stillbirth in this family still concerns me. Matthew's family had a similar history; his grandmother suffered two abortions of male fetuses late in pregnancy. Are these a consequence of Barth syndrome?]

By now, I was beginning to suspect that this disease was not as rare as was generally thought. One possibility was that these children were all members of an extended family, perhaps with just one common ancestor in England. But here science came to our aid: only two of the boys that I have described had the same mutation and already that was being recognised as a "hot spot

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Mutation" [a hot spot mutation is common to many families from different countries, perhaps since this is a particularly damaging site to have a change in the gene], so a common ancestor was not responsible. Also as a result of tests on blood from David's mother and grandmother I was able to explain to Dr. Corner - now a venerable lady in her Nineties - what disease it was that it killed her little patient in 1959.

“The disease can present to haematologists before the onset of heart failure, making it vital to...”

Children Presenting to Haematologists

Still I had it nagging at me that sometimes these children might present to haematologists rather than cardiologists. I spoke to a doctor in the largest specialised neutropenia clinic that I knew of, but this doctor had not heard of Barth syndrome and, when I described it, was confident that no such cases had been seen there. This confidence was misplaced; within a year of that telephone conversation I received a call from a pathologist to say that he had just performed a post mortem on a child who he thought might have Barth syndrome. This little boy had suffered severe feeding problems and during the investigation of these was found to be severely neutropenic and referred to the specialist neutropenia clinic. Numerous tests were performed, except for analysis of organic acids in the urine which would have shown the excess of 3-MGC (this was confirmed later found on a sample taken after death). He was thought to have some form of mitochondrial disease. Granulocyte colony stimulating factor (G-CSF) was commenced to increase his neutrophil count: this shot the count up from less than one to 60, an extraordinary response but one that is not that unusual in Barth syndrome. However he remained unwell with continuing problems with feeding: he became sweaty and breathless whilst taking his bottle and sometimes took hours to feed. This was almost certainly due to worsening cardiac failure although this was not recognised until he developed a serious exacerbation. Despite heroic attempts at his local hospital his cardiac function deteriorated very rapidly and he died.

Sadly, this has not proved an isolated case. I know of two brothers investigated at a specialist haematology clinic for neutropenia and recurrent sepsis without a cause being discerned. One of these subsequently died of heart failure and the other sustained brain damage due to overwhelming sepsis. Both were later recognised as having Barth syndrome. For me, the story of these brothers carries important messages. The disease can present to haematologists before the onset of heart failure, making it vital to make the diagnosis so that heart

function can be monitored and treated as necessary. Secondly, it is incredibly important to recognise the neutropenia and its risks at an early stage so that children can be monitored, treated with antibiotics or G-CSF, as appropriate, in order to try to prevent the more serious forms of sepsis and their potentially devastating consequences. This is particularly so, since the infection problems often seem to reduce with age, in some cases because the neutropenia resolves.

How Frequent is Barth Syndrome?

Space does not allow me to describe all of the children that I see with this disease. However, in the past year alone I have met three other families with this disease, all of whom live in the South of England. This makes me believe that Barth syndrome could be as common as many rare but better-known genetic diseases. For example, I run specialised clinics for children with X-linked adrenoleukodystrophy (the disease described in the film *Lorenzo's Oil*) and Fanconi anaemia. These draw children from a similar catchment area but if I were to run a Barth syndrome clinic this would be of similar size or even larger. I vehemently believe that geneticists, cardiologist, haematologists, neurologists (muscle clinics), metabolic specialists and general paediatricians need to be aware of this disease. Only then can we reduce the risk of death from sepsis and heart failure, and offer carrier detection and antenatal diagnosis most effectively to affected families.

Some Thoughts on The Future

I do not blame doctors - and I would beseech members of this foundation not to either - for failing to recognise this disease rapidly. Even Professor Barth thought it to be extremely rare until recently. It is a sad fact, however, that rare diseases take many years to reach the textbooks and even then will only occupy the small print. Furthermore the escalation of knowledge resulting from modern scientific studies makes it impossible for doctors to effectively keep up with the rate of recognition of new diseases.

I feel that the challenge for modern medicine will be to produce systems which can respond much more rapidly to new knowledge. Health organisations across the world should unite to produce algorithms (flow diagrams) of tests and responses for conditions such as heart failure in children, neutropenia, muscular weakness and failure to

“I vehemently believe that geneticists, cardiologist, haematologists, neurologists (muscle clinics), metabolic specialists and general paediatricians need to be aware of this disease.”

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thrive. These must be free to all users and regularly updated by experts. If, for instance, these included measurement of urinary organic acids and scrutiny of 3-MGC excretion after more routine tests had come up negative, it would be difficult to miss the diagnosis of Barth syndrome.

I do not despair of such systems being produced but it surprises me that they are not already in routine use. Too much of medicine relies on chance meetings and events, anecdote and the use of medical "experts", as I hope my story has made clear. Most months I see children whose diagnosis has been badly delayed for different genetic diseases. In the age of the Internet I feel that it is time for change. By working together with the doctors who treat rare diseases I believe that you, the individuals and families who run self-help organisations like this, can make this wish a reality.

A Postscript

This story has one strange twist: Brandon has never had any abnormality on echocardiography, but his low neutrophil counts persist and have resulted in repeated episodes of skin sepsis and a streptococcal septicaemic episode. His muscles have continued to be weak and he did not walk independently until the age of five years. Despite all his past problems he is now doing well at school. Of all the children that I described he is the only one who does not have a mutation of the tafazzin gene responsible for Barth syndrome. Nor does he have the characteristic abnormality of cardiolipin. So the quest for his diagnosis continues.

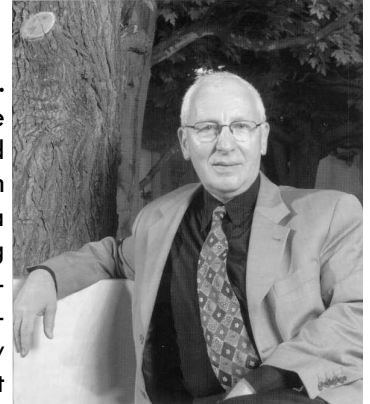
Matthew was a wonderful boy. Sadly, he came to my professional attentions much more seriously five years after his heart transplant. After a period of low lymphocyte counts he developed swollen lymph glands and was shown to have a Non-Hodgkin's lymphoma, a probable complication of the immunosuppressive drugs used to prevent rejection of his transplanted heart. He could not tolerate chemotherapy and died shortly afterwards. I will always remember his bravery and his smiles, and I dedicate this article and my interest in Barth syndrome to him. Anthony remains on drugs for his heart failure six years later and is doing very well.

Acknowledgements

I would also like to thank all of the colleagues who have helped me during the past seven years. Steve Lewis for his inquiring mind (which started it all), Charles Pennock, John Allen and Katie Murdoch-Davies for help with things biochemical, Anthony Oakhill, Nick Goulden and Jackie Cornish (my haematology colleagues) for putting up with my pursuit of this disease, Ann Cantlay for sequencing many genes, Robert Martin and Alison Hayes for help with cardiological aspects of the disease, Michael Ashworth for help with pathology, Philip Jardine for discussions about muscle problems and Ruth Newbury-Ecob for many thoughts on the disease and advice to affected families. ~Colin G. Steward, Ph.D.

IN APPRECIATION OF PROF. PETER J. BARTH

By *Shelley Bowen,*
President



In 1996 I met Prof. Peter Barth for the first time. I had only heard of him by reputation as a result of having two children diagnosed with the disorder that now bears his name. It has always

seemed ironic that such a devastating disorder could be named after this gentle compassionate human being who has devoted his career to medicine to those he cares for. In November I had the honor of speaking on the occasion of Prof. Barth's retirement. Kate McCurdy accompanied me on this journey. Other "Barth" family members attending the symposium were the Stam's, van Loo's, Beemsterboer's, Bleekers, Quant's all from The Netherlands and the Swennen's from Belgium.

We all know the highly valuable contributions Prof. Barth has made toward the scientific advancements in Barth syndrome over the past years but we know little about the many other disorders he has contributed his expertise to during the years of his practice as a pediatric neurologist. It was during this symposium when I more fully realized what a remarkable man Peter Barth truly is. This symposium left me feeling even more thankful to Prof. Barth for his many contributions to science and specifically to the disorder we know as Barth syndrome. He has gone beyond sharing his name with this disorder. When many quality scientists considered it too time consuming, Peter devoted years of dedicated research and attention to those affected by Barth syndrome and other rare disorders. Our children are especially fortunate when we consider just how many other disorders equally deserving are struggling for the attention of talented scientists.

Two-hundred people gathered at this event in honor of Prof. Barth. Prof. H.R. Scholte, the scientist who co-authored the first article about

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IN APPRECIATION OF PROF. PETER J. BARTH

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Barth syndrome in 1983 with Prof. Barth, was present for this symposium. It was a remarkable experience to see these two pioneers in this disorder standing side-by-side. His colleagues Prof. Ronald Wanders, Coby van den Bogart PhD, Peter Vreken PhD†, Bwee Tien Poll-The and Raoul Hennekam were cited by Dr. Barth as being his invaluable partners in science for years. These partnerships have aided not only in the better definition of Barth syndrome but other rare disorders as well.

Prof. Barth acknowledges his wonderful wife Hetty as an instrumental influence in his career. I can fully understand how she would be a positive influence on all those around her. She is tender in spirit, intelligent in mind and gracious in personality. She and Peter Barth have two children, Serge and Trudi who were both present as well as Trudi's husband Ron Vandersteen and Prof. Barth's brother Michael and his wife Ellen.

Among the notable speakers were Dr. Leo Smit, President of the Dutch Pediatric Neurology Society who opened the symposium noting his longtime work with Prof. Barth and Prof. Barth's role in creating The Dutch Pediatric Neurology Society. Richard Kelley, M.D., Ph.D., Division of Metabolism, Kennedy Krieger Institute, Department of Pediatrics Johns Hopkins Medical Institution; Eugen Boltshauser, M.D., PhD, Department of Pediatric Neurology, University Children's Hospital Zurich; Harvey Sarnat, M.D., F.R.C.P.C. Departments of Pediatrics (Neurology) and Pathology (Neuropathology), Cedars-Sinai Medical Center and UCLA School of Medicine, Los Angeles, California, U.S.A; Raoul CM Hennekam, M.D. Ph.D., Departments of Pediatrics and Clinical Genetics, AMC, Amsterdam; Prof. Bwee Tien Poll-The, M.D, Ph.D. AMC, Pediatric Neurology and Prof. Barth's successor; Ronald Wanders, Ph.D.

It was moving to hear Dr. Barth speak about the two types of disorders he has spent his professional career investigating, specifically, Fetal brain disorders and Neuromuscular disorders. His lecture was aptly titled "The challenge in hidden treasures-a never ending story". In addition to his scientific deliver he spoke to scientists in the audience about serendipity and advised the young scientists in the audience to always be on guard for this because one never knows when serendipity may occur and change the course of scientific understanding of diseases.

Prof. Hugo S.A. Heijmans ended the symposia with a delightful presentation of Prof. Barth's dedication throughout the years including many personal photographs such as a young Peter Barth as a child, a wedding photo of Hetty and Peter Barth, a loving photo of Peter Barth as a father and one as a physician with a patient, obtaining his professorship, and with his beloved parrot. His dedicated service as a teacher and physician was clearly highlighted.

During a reception hosted by AMC all of the BSF families present had the privilege to meet with Annalies Vreken and personally extend our appreciation of her late husband Peter's dedicated research. His uniquely generous spirit and research added much to the understanding of this disorder. His dedication was admired and respected by all who knew him. At the end of the evening it was clear to see the world of science has been deeply enriched by Peter Barth's invaluable dedication and years of medical expertise. It is also important to note that Prof. Barth will continue to work on the specific disorders he has researched over the years and will remain with BSF for some time to come. This occasion marks a time for him to focus more deeply on the complexities of the disorders on which he clearly qualifies as "expert" physician and teacher. We are fortunate to be the benefactor of his knowledge and continued research.

~Shelley Bowen

To view pictures of Dr. Peter Barth's Symposium, please visit www.barthsyndrome.org/barth_retirement/index.html



Front row from left to right: Prof. Harvey Sarnat (LA), Prof. Leo Smit (Ped. Neurologist, Free Univ. Med. Ctr. Amsterdam), Prof. Peter G. Barth, Mrs. Hetty Barth, Shelley Bowen, Prof. Bwee Tien Poll-The (Ped. Neurologist Academic Med. Ctr. Amsterdam and Prof. Barth's successor) and Prof. Eugen Boltshauser (Univ. Children's Hospital, Dept. of Ped. Neurology, Zurich Switzerland).

Back row from left to right: Dr. Richard Kelley (Baltimore), Dr. Raoul Hennekam (Pediatrician/Clinical Geneticist AMC Amsterdam) and Prof. Ron Wanders (Laboratory of Genetic Metabolic Diseases, AMC Amsterdam)



CARDIOLIPIN TAKES CENTER STAGE

By Miriam L. Greenberg, Ph.D.

Ask people to conjure up an image of a scientist at work, and most will describe a person (usually male) in a white coat staring at a test tube, thinking esoteric thoughts and feeling very pleased as new insights just rain down on him. The reality for this (female) scientist is better described by 11 early years of continual failure to clone the yeast genes that code for the cardiolipin biosynthetic enzymes. I look back on those years as the 'head banging' period. Of course, not all my time was spent head banging; like all scientists, I devoted a great deal of effort to writing grant applications to fund this frustrating work. Far from esoteric, my thoughts usually ran to "Still trying? You should have your head examined!" Finally, in 1997, a very talented graduate student in my laboratory, Feng Jiang, succeeded in identifying the cardiolipin synthase gene. Because yeast is a very tractable model organism, he was able to make a "knock-out" yeast cell that lacked the gene and, thus, could not make cardiolipin. This work finally enabled us to ask important questions about cardiolipin, i.e., what happens when cells don't have cardiolipin, and how is cardiolipin synthesis regulated? Far from head banging, the last 6 years have been very exciting as we gain insight into these questions.

"More important, however, this was the first time that a direct association had been made between cardiolipin and a specific human syndrome."

I first heard about Barth syndrome in the fall of 2000, while I was on sabbatical in Utrecht University in the Netherlands. Michael Schlame wrote urging me to contact a scientist by the name of Peter Vreken in Amsterdam, who had just discovered that cells from patients with Barth syndrome could not properly remodel cardiolipin. (Cardiolipin remodeling is the process whereby the fatty acids that are present on a cardiolipin molecule are replaced with different fatty acids). I immediately contacted Peter, who generously invited me to Amsterdam to meet with him and his colleagues, and to discuss the possibility of a scientific collaboration. This meeting was very inspirational for me. For one thing, Peter's group had constructed a knock-out yeast cell (a *taz1* mutant) that lacked the Barth syndrome gene. Therefore, it was now possible to study cardiolipin remodeling in yeast. More important, however, this was the first time that a direct association had been made between cardiolipin and a specific human syndrome. Needless to say, this link has been a powerful motivating factor in our work.

The prospect of presenting my work at the Barth Syndrome Foundation meeting in Baltimore was very exciting. I had never once attended a scientific meeting in

which cardiolipin took center stage, or was even the topic of a major session. Almost all my colleagues in the field, some of whom I had not seen in years, would be attending the meeting. In addition, I very much looked forward to finding out more about Barth syndrome, and the role of cardiolipin in this illness. I am pleased to summarize the key findings presented at the "Biochemistry of Cardiolipin" and "Biochemistry of Barth Syndrome" sessions of the meeting, and to share my overall impressions of this outstanding meeting.

"Could it be that cardiolipin did not play an important role in cell function? A resounding no is the answer to this question."

As the first speaker of the session, I briefly summarized early studies carried out in many laboratories all suggesting that cardiolipin is required for the function of many mitochondrial enzymes. These early studies were performed in vitro, i.e., "in the test tube." To do so, mitochondrial enzymes were isolated from cells, and their activities were measured in the presence or absence of cardiolipin. While these studies strongly suggested an important role for cardiolipin, this role had to be tested in vivo, i.e., in a living cell. In vivo studies to look at the role of cardiolipin awaited the availability of an organism in which cardiolipin was deficient. Our yeast cardiolipin synthase knockout mutant, *crd1*, provided just such an organism. The *crd1* mutant cannot make cardiolipin. Therefore, we can do experiments to identify mitochondrial functions that are defective in the cardiolipin-deficient cell, when compared to the wild type control cell. To summarize our findings, while a number of enzymatic activities are reduced in the cardiolipin-deficient mutant, cardiolipin is not required for respiration during optimal conditions! This was initially surprising in view of the in vitro experiments identifying so many respiratory enzymes that seemed to require cardiolipin for activity. Could it be that cardiolipin did not play an important role in cell function? A resounding no is the answer to this question. We observed that cardiolipin-deficient cells have a clear growth defect at elevated temperatures. Furthermore, while mitochondrial function seems fine under ideal growth conditions, it turns out that cardiolipin is required for mitochondrial function under stressful conditions. For example, oxidative phosphorylation is completely uncoupled in cardiolipin-deficient mitochondria at elevated temperatures. Uncoupling is also more apparent in mutant mitochondria when the rate of respiration is fast. Finally, cardiolipin-deficient mitochondria cannot swell and shrink appropriately in response to osmotic stress. In

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CARDIOLIPIN TAKES CENTER STAGE

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short, during ideal growth conditions, the absence of cardiolipin does not lead to obvious cellular defects. However, cells lacking cardiolipin cannot respond appropriately to deleterious conditions such as elevated temperature, osmotic stress, and fast respiration. Does cardiolipin remodeling play a role in the ability of cells to respond to these stresses? We have begun to address this question using the yeast *taz1* mutant.

Two very exciting talks on cardiolipin remodeling followed. University of Manitoba professor Grant Hatch is an expert in the field of cardiolipin remodeling in mammalian cells. His work has been pivotal in identifying conditions that regulate cardiolipin synthesis and remodeling in the mammalian heart. He has identified four physiological factors that affect cardiolipin synthesis, including substrate availability, thyroid hormone, ceramide signaling, and phosphorylation. The fact that cardiolipin synthesis is so highly regulated supports the view that this molecule plays an important role in heart function. In addition to regulation of cardiolipin synthesis, Grant Hatch's laboratory has contributed

“The most exciting recent work in the Hatch laboratory may have far-reaching implications in elucidating the function of the TAZ1 gene.”

greatly to our understanding of how cardiolipin is remodeled. Because remodeling involves the replacement of fatty acids in cardiolipin with new, mostly unsaturated fatty acids, he hypothesized that import of fatty acids into the mitochondrion may play a role in regulation of the remodeling process. However, this proved not to be the case, as inhibition of fatty acid import does not affect remodeling of cardiolipin or the precursor lipid phosphatidylglycerol. The most exciting recent work in the Hatch laboratory may have far-reaching implications in elucidating the function of the TAZ1 gene. This is the development of an *in vivo* system in which to characterize the protein encoded by TAZ1. In this system, the TAZ1 gene from mouse liver was successfully over-expressed in a COS cell line. This was demonstrated by increased incorporation of the unsaturated fatty acid linoleic acid into cardiolipin in cells over-expressing the TAZ1 gene. This tool is a very powerful system that offers tremendous promise for elucidating the mechanism and function of cardiolipin remodeling.

Dr. Michael Schlame (NYU) followed with an exciting talk describing cardiolipin-related abnormalities in Barth syndrome cells. Dr. Schlame has carried out fundamental studies of cardiolipin biosynthesis in mammalian cells over the course of the last 12 years. In addition, he developed a sensitive biochemical technique to measure the different acyl species of cardiolipin, which

he has applied to the study of cardiolipin defects in Barth syndrome. Recently, Dr. Schlame has shown that Barth syndrome cells have an abnormal cardiolipin composition. Specifically, the species known as L4 (in which all 4 acyl groups are linoleic acid) is severely deficient in platelets, skeletal muscle, heart, and fibroblasts. Interestingly, he did not observe a correlation between clinical severity and the degree of L4 deficiency. However, L4 deficiency appears to be specific

for Barth syndrome and was not observed in other myopathies. What is the function of L4? One clue is apparent in the correlation that tissues enriched in L4 are those that have a greater degree of mitochondrial structure, such as heart and skeletal muscle, which require mitochondria to make ATP during exercise. Liver cells, in contrast, have a lower amount of L4. Understanding the specific function of L4 will no doubt have important implications for our understanding of the defect in Barth syndrome. In addition to the identification of L4 deficiency, Dr. Schlame has made very important observations that have direct bearing on the mechanism of cardiolipin remodeling. There are two potential

sources of fatty acids for remodeling, free fatty acids (acyl CoA-dependent acylation) and phospholipids (acyl CoA-independent transacylation). Interestingly, the levels of CoA-dependent acylation are increased in Barth syndrome. Even more exciting is the observation that incorporation of linoleic acid into cardiolipin appears to be acyl-CoA independent, suggesting that the fatty acids are derived from other phospholipids via transacylation. This mechanism appears to be specific for acylation of cardiolipin with linoleic acid (18:2), and the donor lipids are either phosphatidylcholine or phosphatidylethanolamine. These very novel findings cause us to completely re-think our current model (which was actually proposed by Dr. Schlame) for cardiolipin remodeling, and will no doubt help to provide crucial insight into the defect in Barth syndrome.

While we all seem to focus our studies on mitochondrial defects in Barth syndrome, Dr. Richard Kelley (Johns Hopkins Medical Institutions) pointed out that Barth syndrome does not present with symptoms and pathologies that are typical of mitochondrial diseases. For example, muscle biopsies appear to be normal in some Barth syndrome patients. On the other hand, growth retardation is often out of proportion to the mitochondrial deficiency observed in Barth syndrome. Consistent with this observation, the G4.5-encoded protein may

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CARDIOLIPIN TAKES CENTER STAGE

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localize to the plasma membrane. Finally, muscle cells from Barth syndrome patients exhibit normal ATP bioenergetics, as do fibroblasts. Clearly, we have yet to understand how a defect in mitochondrial lipids can explain these diverse phenotypes.

The session closed with a talk by Troy Phipps (University of Southern California), who has been carrying out experiments to determine the subcellular localization of the G4.5 protein in Barth syndrome lymphoblasts immortalized with EB-virus. Mr. Phipps' data suggest that the protein may be localized to different regions of the cell in addition to mitochondria. As such, it is possible that a reduction in cardiolipin remodeling may not be the only defect that could account for the pathophysiology of Barth Syndrome. Mr. Phipps indicated that one problem with these studies is that the antibody used is not highly specific for G4.5. Therefore, these observations must be validated with more specific antibodies.

“The predominant tone at this meeting was one of collaboration and free sharing of knowledge. We are all in debt to Peter Barth, whose insights and scientific generosity continue to guide our research in this field.”

I will end this summary with some personal observations about the meeting. I believe that the tone of a meeting owes a great deal to the leaders in the field. The predominant tone at this meeting was one of collaboration and free sharing of knowledge. We are all in debt to Peter Barth, whose insights and scientific generosity continue to guide our research in this field. In addition, the spirit of the late Peter Vreken was present throughout. Peter made the crucial observation that cardiolipin content and acylation are defective in Barth syndrome. It was this finding that inspired virtually all of the work presented at this meeting. While we miss him as a colleague, his contribution is very strongly felt. Finally, I would like to add that during

all the years of frustration with experiments that I described above, it was almost impossible not to wonder if there was some point to pursuing this project. However, I was inspired by the tremendous strength of the families of all the beautiful boys at this meeting. If our studies of cardiolipin function can shed some light on Barth syndrome, then all the early failures will certainly have been worthwhile.

~Miriam L. Greenberg, Ph.D.



A COLLABORATION OF NEW IDEAS

“This broad cross-section of investigators were able to teach each other the current status in their respective areas...”

The Barth Syndrome International Family & Scientific Conference, co-sponsored by Office of Rare Diseases (NIH), National Institute of Neurological Disorders & Stroke (NIH), and the Lied Foundation Trust, Christina M. Hixson, Trustee, was held on October 18-20, 2002 in Baltimore, Maryland. There is a consensus in the air that this conference was a huge success on many levels. “It was exciting to see support from NIH (ORD and NINDS) for this rare disorder,” stated Dr. Ariel Sherbany, a Nanuet-based pediatric neurologist who has cared for a patient diagnosed with Barth syndrome for many years.

Benefits of Combining Clinical/Scientific Presentations

“The excellent organizational plan and attention to detail was first rate and the medical-scientific participants were both expert and involved,” stated Dr. Jeff Towbin, Associate Chief Pediatric Cardiologist at the Department of Pediatrics and Molecular and Human Genetics, Baylor College of Medicine, Houston, Texas. “As a participant, it was a ‘breath of fresh air’. The clinical/scientific presentations were excellent and combined clinical cardiology, hematology, and neurology with basic genetics, biochemistry, and mitochondrial bioenergetics and gene therapy, amongst others. This broad cross-section of investigators were able to teach each other the current status in their respective areas and also conveyed these exciting advances effectively to an interested group of “lay” people, the families. This is unusual and difficult to master; however, it worked!”

“The community of researchers interested in Barth syndrome is small and most scientists have known each other from publications,” said Dr. Michael Schlame, Department of Anesthesiology, NYU School of Medicine, NY, NY. “However, it was at the conference where scientists actually met for the first time and there is no doubt that the meeting invigorated new collaborative projects. It was in my conversations with several physicians and scientists that I realized they invariably praised the scope and quality of the scientific sessions and expressed confidence that the conference will have a positive impact on research activities related to Barth syndrome. Having had the chance to discuss various aspects of Barth syndrome with experts was extremely important to me. A few highlights of new information I walked away with include the fact that (1) Barth syndrome has variable clinical expressions, and that it is most important to recognize the disease early in life (appropriate management of the specific cardiologic and

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KNOWLEDGE AND INSIGHTS SHARED DURING YOUTH FORUM

By Jaclyn Butera, MSW, CSW, Med

(Disclaimer to the parents and to the "Barth Boys": If I appear vague in this article, it is in the name of wanting the boys to feel as if our private conversations and sharing will remain "top secret", to be disclosed only at their own discretion and to their chosen audience. As I write I ask parents to respect the individuality and confidentiality of each of the boys. And to the boys who will read this, I must say this-you have indelibly left your mark on my heart and in my mind. Please... keep painting your rocks and don't worry about naming them! -JB)

Having worked with youth in various capacities, case worker, youth advocate, dean of student life and academic dean and clinician, I thought the idea of running a group especially for the "Barth Boys" was a fantastic opportunity on so many levels. There certainly seems to be a natural inclination for individuals to gather in groups for mutually beneficial purposes and reasons, and despite the notion that this group is united largely by struggle and challenge, I knew from my school contact with one of the boys that those beneficial purposes would be at the center of our shared work. I was right. What an inspirational group of boys and young men! In our pre-group meeting we got acquainted and worked to establish some level of comfort and trust, and to formulate questions and statements of concern for the panel discussion.

It was a genuine privilege to work with such a dynamic group of individuals. This much became immediately clear: each one brings his own special gift to the table. While the boys are obviously joined by some common experiences, they are ultimately individuals. Each one lives his life, encounters his own joys and challenges, and ends each day with his own sense of himself.

Schools can be such wonderful environments, and I'd venture a guess that many adults, removed from school experiences by at least a handful of years, look back fondly on the lessons learned. What we sometimes lose sight of is the fact that each and every day at school is a new experience, one that can be tremendously uplifting, fulfilling, and positive, or it can be sad, discouraging, and painful. The sharing that these boys offered of themselves and to one another is so beneficial, for it helps to reestablish/underscore their common bond while it gives them perspective for processing those more difficult times. The boys, by being generous with one another and by absorbing what was shared, left the conference with a sense that they are "normal" in a great many important ways. Peers who might want to help may turn away out of discomfort; those who aren't as sensitive



Will McCurdy, Jackie Butera and John Wilkins

might be shortsighted, cruel, or dismissive. In this group, however, there is something that replaces one norm with another. It was my observation that this kind of sharing is so beneficial for these boys. They create a "look at me - I am normal" mentality, one that helps to normalize their experiences more than anyone else can.

"A common theme...was that all of the boys have, at one time or another, felt bullied."

Understanding is the product of wisdom, and some school-age young people are not as open and accepting as they might be. A common theme that emerged during our pre-group conversations was that all of the boys have, at one time or another, felt bullied. We know from countless sources that students who are the target of bullying commonly suffer academic, physical, and emotional consequences, and while this part of our sharing was certainly less uplifting, it was no less important or potentially important. In the sharing we recognized that what happens is not right, regardless of how many voices in the community might say, "Kids will be kids." Please do not allow the school environment to minimize the effects of this kind of targeting on your boys. Schools should never minimize, tolerate, or ignore the extent of bullying and the harm it can cause. Recent reports on bullying point out that victims often have difficulty concentrating on schoolwork, show low performance, and have relatively high rates of absenteeism. Some of the emotional symptoms or signs that bullying may be occurring include anxiety, depression, and low self-esteem. To counteract bullying, schools

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A COLLABORATION OF NEW IDEAS

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hematologic problems from the first days of life may result in excellent outcome); (2) Three laboratory tests can help to establish the diagnosis of Barth syndrome, that being (a) sequencing of the TAZ gene; (b) analysis of cardiolipin in platelets, and (c) the measurement of organic acids in urine; (3) There are patients who present with the characteristic features of Barth syndrome, yet who have a normal TAZ gene and normal cardiolipin in platelets - this suggests that Barth syndrome may be caused by more than one genetic defect; (4) There is exciting new research under way to elucidate the physiologic mechanism of Barth syndrome; and (5) The current treatment of Barth syndrome is symptomatic; however, new therapeutic approaches using dietary modulation were discussed at the meeting.”

“Dr. Iris Gonzalez’s genetic and DNA studies were fascinating and tantalizing in so far as they promise to unlock the mystery...”

Dr. Sherbany, who had the privilege of attending both the June 2000 and October 2002 Barth Syndrome conferences, stated: “The Barth Syndrome Conference was a very exciting place to be. What was most delightful was that it was well attended by patients, doctors and scientists. It was clear that progress has been made. With a disciplined approach by scientists like Dr. Peter Barth, Dr. Richard Kelley and many others who are committed to

finding out the underlying etiology of this disorder our hope of finding a cure and treatments to ameliorate this condition have been advanced.” Dr. Sherbany added: “It is frustrating that, although patients with this disorder are living longer with a better quality of life, we still do not know enough and we still have a long way to go.”

When asked to summarize the highlights of the conference, Dr. Sherbany stated: “Dr. Iris Gonzalez’s genetic and DNA studies were fascinating and tantalizing in so far as they promise to unlock the mystery of the gene product that may be responsible for this disease. Another breakthrough may be the research presented that seems to tie cardiolipin deficiency to this disease. Other topics of interest included the issue of under-diagnosis of this disorder and the differences in treating neutropenia in Barth syndrome (i.e. perhaps requiring low dose GCSF). The review of the cardiac, neurological, and metabolic issues were also very interesting and helpful to the diagnostic and treatment issues. It is apparent from the DNA studies, as well as the clinical manifestations, that although this is a rare disorder the manifestations are pleomorphic with different degrees of severity. As patients are living longer with improved cardiac care and care of neutropenia, we may be seeing age-dependent

variations and exacerbations of symptoms. Because it is a rare disorder it may be hard at times for the clinician to determine whether symptoms like headaches, chronic fatigue, etc. are due to the disease or some other unrelated etiology. It is a conference of this sort and a registry of patients that will eventually help in sorting these issues out. The work by these scientists can have relevance to other metabolic disorders as well and may also uncover understanding of cardiolipin metabolism as it relates to body functions. What does cardiolipin do? Why do we need it?”

How and why did the interweaving of families, scientists and clinicians occur?

“Clearly, the organizers understand the needs of the boys and their families,” stated Dr. Towbin. Impressively, they also understand the current state of the literature, the clinicians and scientists responsible for the ongoing investigations, and conveyed the need for direct interaction between these diverse parties. Again, it worked! The concept allowed direct discussions between families, scientists and clinicians. The enthusiasm of the families was infectious and created an atmosphere of caring and intellectual pursuit intertwined. My hat is off to the organizers. I appreciated the opportunity to participate and hope the future brings new discovery, better treatments, and answers to the questions that you all have.”

“The combination of the scientific and family meetings contributed to the success of the conference,” said Dr. Schlame. “This was the unanimous opinion of all participants. All found it very motivating to meet children with Barth syndrome and their families. It helped to put research into perspective and it was a great human experience.”

“It was heartening and touching to meet some of the Barth syndrome families and children and see them get together and be supportive of each other,” said Dr. Sherbany. The conference offered an opportunity to mix with some of the scientists and physicians and bat some ideas between them. It was a very important opportunity to baseline the patients and the clinical spectrum of the disease as a potential for future studies that may help improve treatment and find a cure for this disorder. The conference embodies the response and commitment by the Barth syndrome families that is very reminiscent of the Lorenzo’s Oil story. As an aside, we were privileged by the presence of Dr. Hugo Moser at this conference, who has contributed immensely to the understanding of the Lorenzo’s oil disease. This type of approach to rare disorders can serve as a model for solving other rare disorders.”

~Lynda Sedefian



EXCITING SCIENTIFIC AND MEDICAL ADVISORY BOARD NEWS

By Kate McCurdy, Vice President, Science and Medicine

All of us associated with the Barth Syndrome Foundation have much to be proud of and even more to be grateful for. Our Scientific and Medical Advisory Board (SMAB) falls into both lists! The twelve physicians and research scientists who comprise this board provided the backbone of information at our recent conference in Baltimore. *Every single one* of them was with us for at least two days and was involved with a presentation (or several) at the Family or Scientific meetings, and many also participated in the various clinical sessions that were held with individual families. Furthermore, some were helpful in identifying and recruiting additional speakers who were of equally high quality and interest. We are *extremely* appreciative of the time, effort and expertise the SMAB members put into making the conference so successful -- particularly Dr. Richard Kelley, the Chairman of the SMAB and our host in Baltimore.

I mentioned that there are now twelve professional members of the SMAB. That is one more than we had the last time this group was written up, since we are fortunate that Dr. Barry J. Byrne agreed to join the team in September 2002. Dr. Byrne is a pediatric cardiologist at Shands Children's Hospital in Gainesville, FL, where he cares for two Barth boys, and a Professor of both Pediatrics as well as Molecular Genetics and Microbiology at the University of Florida School of Medicine. He is Director of the Powell Gene Therapy Center and recently has been working on the development of gene replacement possibilities for Pompe's Disease. Those who were at the Baltimore conference in October will remember Dr. Byrne as an entertaining and informative speaker, a gentle and caring physician and a very involved member of the group. Welcome to our midst!

As I write this article, another really important milestone for BSF is being achieved with the help of our SMAB. We announced last spring that the Barth Syndrome Foundation, Inc. was launching a research grant program to fund research "on the natural history, biochemical basis, and treatment of Barth syndrome." We set out to provide "'seed money' to be used by experienced investigators for the testing of initial hypotheses and collection of preliminary data leading to successful long-term funding by NIH and other major granting institutions. In addition, we [have particular interest] in attracting new investigators to the very interesting field of Barth syndrome research." We created a two-stage grant process whereby Letters of Intent were due to us

by the end of June 2002; we sent out almost 2,300 pieces of mail to medical centers and hospitals; we launched an e-mail campaign; and Shelia Mann and Steve Kugelman helped us post information about our program on various internet sites. Once received and reviewed by our SMAB for scientific merit, the BSF Board of Directors invited seven Principal Investigators to submit full research grant applications to us by October 15, 2002. We received all of these just before our conference, and I must say that they are of absolutely top-notch quality, from highly qualified individuals at world class institutions in four countries. They represent a variety of interesting investigations about topics critical to the advancement of knowledge and treatment of Barth syndrome. No organization could have hoped for more! And especially not one that was just beginning such a program. These grant applications are currently being evaluated by the non-applicant members of our SMAB as well as several outside reviewers. Armed with recommendations from this group, the BSF Board of Directors will announce the final awards on December 9, 2002. It will be an extremely exciting moment for us all when some of the funds that have been raised through all the efforts and activities of various members of the Barth Syndrome Foundation are directly applied to high quality research specifically pertaining to the disorder. Congratulations one and all!

~Kate McCurdy



ANNOUNCING:

THE BARTH SYNDROME FOUNDATION 2003

GRANT RESEARCH PROGRAM SCHEDULE

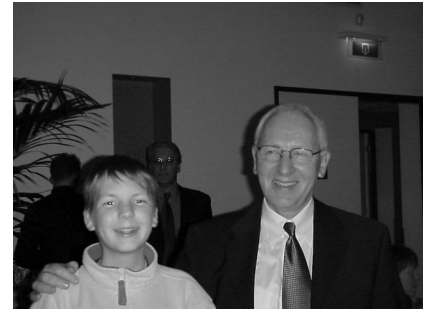
June 27, 2003—Deadline for Receipt of Letter of Intent by BSF

July 30, 2003—Notification date for writers of Letters of Intent

October 15, 2003—Deadline for receipt of full grant applications by BSF

December 5, 2003—Notification date for grant applicants, with funding to begin soon thereafter

For further details, please visit the BSF website at www.barthsyndrome.org sometime after January 1, 2003



Peter van Loo, who had the honor of being present at Dr. Barth's Retirement Ceremony, presented a 'scrapbook' to Dr. Barth on behalf of the Barth syndrome families.

Dear Families,

On the occasion of my official retirement from my position at Amsterdam University and Emma Children's Hospital a symposium was organized with many attendants and speakers, including a representation from B.S.F. by Shelley Bowen, Richard Kelley and Kate McCurdy. I was surprised and pleased to receive on behalf of the Barth syndrome families an impressive scrapbook with so many endearing and heartening contributions.

Because it is not possible to thank each of you in person, I would like to thank you all by making use of this Newsletter. Beside the presence of so many, it was the most precious gift that I received on this occasion. On behalf of myself, my wife Hetty and children, thank you so much!

Many asked what I would be doing next - eh, fishing? No! Although you never can be involved for more than a lifetime, I will be involved in the work of the B.S.F., including the European branch of the network, and I will be available, as long as I can contribute essentially to our common aim. Also my work at Emma Children's hospital will go on, from now on a part-time basis so that we can stay in touch. As a line of information for all of you: just as before, Barth syndrome will remain a major focus of research at Amsterdam University and Emma Children's hospital, lead by Professor Ronald Wanders. My official successor to the Chair of Pediatric Neurology is Prof. Bwe Tien Poll-The; her major interest is in inborn errors of metabolism. We worked closely together and will go on to do so.

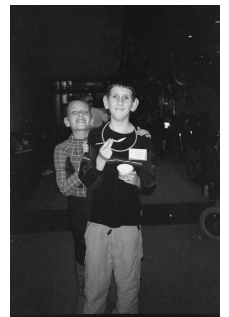
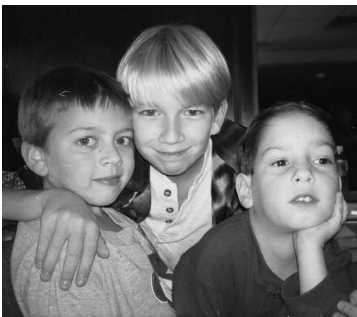
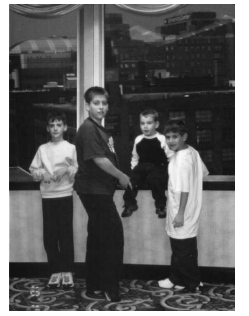
May I wish, to all of you, a nice Christmas and a beautiful year to come!

~Peter G. Barth



The docs enjoy some down-time at BSF's Saturday Night Social Event which took place at the Turner Lobby, Johns Hopkins University School of Medicine. This event was co-sponsored by Starlight Children's Foundation—midatlantic and A.C. and Rosa Mann of Chattanooga, TN. From left to right: Miriam Greenberg, Iris Gonzalez, Barry Byrne, Ariel Sherbany, Ruth Newbury-Cobb, Colin Steward, Jules Spotts, Gerry Cox, Troy Phipps, Joan Stoner, Eileen Juico, Jackie Butera. (Special thanks to Cara Spallone, Elaine Stashinko, and Glenn Comia for their generous assistance.)

BSF's SATURDAY NIGHT SOCIAL EVENT



KNOWLEDGE AND INSIGHTS SHARED DURING YOUTH FORUM

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can distribute a written anti-bullying policy that is applied consistently and fairly to everyone in the school community. In my opinion there are enough issues at hand not only for these remarkable boys but also for all students. That we are all vigilant in protecting students' rights is essential.



A Time to Reflect ~ Michael Bowen

Our discussion with the doctors was another great opportunity, one that was appreciated by both the doctors and the boys. The incredible breadth and depth of knowl-

edge and insights that the doctors were able to offer was so obviously valuable to the boys. As familiar as they may be with the ins and outs of their personal situations, the boys clearly benefit from the information shared by the doctors. Their connection to each other and to a variety of supports is a crucial need.

Perhaps the most significant moment came when the doctors offered information that was reassuring; every one of the boys was pleased and encouraged to hear that there are men living with Barth Syndrome who lead full lives and who have children. Will McCurdy had this to say about his session with the doctors: "The session with the doctors was one of the highlights of the entire conference for me. It was wonderful to be able to sit down with the world experts and have questions that were important to us answered. I think that it is one of the things that the Barth Syndrome Foundation is striving for, getting the tough questions answered so people can be empowered by that knowledge. I am very grateful for the interest the doctors have shown in our cause." Will's combination of honesty about having his concerns addressed and gratitude that he has access to the best of those answers is indicative of the positives that came out of the conference.

As I completed my role at the conference, I reflected on what I had observed and thought about what suggestions I might make so that the Foundation can continue to support these boys in substantive ways. Clearly, there is a need for activities and interactions that continue to bolster the boys' sense of self worth and self-esteem. I wonder if there aren't technological means, or camp programs that could be designed specifically for maintaining contact through the coming years. Not only will this add strength but it will serve to decrease the sense of isolation that certainly accompanies returning to a

home and a school setting in which the commonality of experience, which was so rich and vital and important during our days in Baltimore, is not readily at hand.

Another thought centered on the notion that, as much as it was easy to consider the boys as a group, as a unified collection, it is important to remember that each boy is at a different stage of personal development. This reminds us all that each boy will hear and process information about health, the future, and any of a thousand other concerns, in different ways. And as they continue to develop, they will manage this information differently every time they hear it. So, there is a constant and ongoing need to provide them with the opportunity to get answer to questions.

It's important that we are diligent in keeping these boys as connected as possible to their communities and schools in order to minimize isolation. The bullying issue under-

"It's important that we are diligent in keeping these boys as connected as possible to their communities and schools..."

scored the importance of this need, even if this is not the central issue in the life of each boy. Conditions permitting, the boys spend a good percentage of their time in school, and we must do what we can to educate schools, to work with school personnel, and to be sure that all parties are sensitive to the evolution of each boy's situation. My own experience has suggested that schools can be wonderfully responsive and accommodating, and it has also shown me that schools can make assumptions. Those of us in schools must reevaluate and reassess our

supports, even in the face of this week's successes so that next week's plans can be fruitful. In order to offer the best possible support and service, we must work closely with the student and his family, anticipating when and where we can and reacting as best we can.

In many ways children are the best teachers for other children. As much as I was honored to bring a professional expertise to the conference, I was humbled and impressed by the interactions that we established as a group but that were carried out in substance by the boys. Listening to the older boys/young men offering insights and encouragement to the younger ones in the group suggested that what I had helped to initiate was really an opportunity for the real experts to share with each other. I look forward to maintaining my own contacts with the extraordinary group I met in Baltimore. I still have a lot to learn from them. Please feel free to contact me at Jacquie_Butera@rcds.rye.ny.us or by phone at (914) 925-4636. ~Jaclyn M. Butera, MSW



CHANNELING THE REALIZATION OF POWER AND SENSE OF INDIVIDUALITY

By Jules Spotts, Ph.D., P.C.

During the recent conference in Baltimore, several parent sessions were held, one for parents of younger boys with Barth syndrome and one for parents of older boys with Barth Syndrome. I was fortunate enough to be part of both panels. It is my intent to summarize the major points which emerged in these two meetings - they bear repeating.

Basic to every boy with Barth syndrome is the notion that he is an individual, a male, who has Barth syndrome as one characteristic of who he is. The definition of self by each of our boys does not have as its' central component Barth syndrome. In much the same way as someone who suffers from diabetes has the task to not make that the defining factor of self, **boys with Barth syndrome are not defined by their illness.**

A second factor of vital importance is the need to help boys with Barth syndrome to explore and develop a variety of interests as we help them to find ways to have channels for social entry. Although there are physical limits present, the boys can explore and expand strengths which they do possess. The additional positive result of this can, in fact, be the realization of "power" which they need to experience. The more we can help them to develop those strengths, the more they will find a sense of power over the syndrome.



Jules Spotts, Ph.D., P.C.



This handsome group of boys and young men united in Baltimore to be with their friends—friends many have not seen since the last conference in 2000. Here they stand proud as if to say: "we are individuals first, with many different components of our individuality.

lescent years. As kids develop beyond that early adolescent stage, the range of groups and ways in which to belong will only increase, thus, helping our population immensely. Starting early with our group of boys around self awareness and potency within limits, each boy's ability to the development of a diverse personality, will be of value.

As I continue my involvement with Barth syndrome families, I continue to be impressed with the positive force generated by everyone. If we adults, parents and professionals, continue our commitment to active coping with Barth syndrome, further research and discovery and eventual cure, we set wonderful examples for the boys themselves, who ultimately get the lion's share of the credit for their force in succeeding to develop and indeed to thrive.

~Jules Spotts, Ph.D., P.C.



BSF OFFERS NEW HOPE AND A SENSE OF BELONGING

One week prior to BSF's 2002 International Family and Scientific Conference, President Shelley Bowen received two phone calls from two different families who had either **just** received a positive diagnosis, or were awaiting a diagnosis for their son.

We regret to report that Aden Edsel, 11 months, died from cardiac failure on November 23, 2002.



Holly and Aden Edsel†

Prior to his death, Aden's mom, Holly, was asked to write an article for this newsletter. When asked whether she would still like to have her article included, Holly responded: "Yes, Aden was here for a purpose. Perhaps the research into Aden's death will help other boys dealing with this disorder. I chose this picture of Aden and I, which was taken on our way to BSF's 2002 conference because it resembles the happiest I have ever seen Aden. It was as if Aden was guiding me to a place where *he knew* I would receive the answers that I so desperately needed." Holly's letter clearly expresses the emotions we have all felt, especially the realization that we are no longer alone.

For Holly Edsel, her journey began on October 9th when her son Aden was in the hospital for the second time in less than two months for cardiomyopathy and Rotto Virus, yet she still had no knowledge of why Aden's heart was failing. "Our cardi-

ologist said it was idiopathic and we would probably never know, but thank God the geneticist did not agree" said Holly. She was told that the geneticist would be in touch within the next two weeks...that he wanted to rule out the possibility of any syndrome. One hour before Aden was discharged from the hospital their geneticist told Holly that Aden had Barth syndrome. "I then felt a sigh of relief—no more guessing games," said Holly. "I now asked myself 'What could I do as a mom to help with this?'" Holly decided to turn to the Internet where she found BSF's website and read about the upcoming conference, and, "the next thing I knew my mother, Aden and I were on a plane to Baltimore with very little information and a brand new diagnosis."

Upon arrival at the Holiday Inn in Baltimore, Moira Masterson, a grandparent whom Holly's mom had corresponded with via email, met them in the lobby. "My mother just glowed as she knew immediately that we had done the right thing...she knew that even though this was the first time she had ever seen this woman that they were going to be great friends."

Holly mentioned that she met many families, shook many hands and hugged many moms that were at the reception. "In my heart I knew I was where I belonged. For some reason God had brought this loophole into my life and maybe it was for a reason, but I knew it had to be a good one. Aden is sick, as all of our children, but he is so special that I can't put into words how it feels to know that I have the privilege of being his mother."

"The conference was the most wonderful thing that has happened since the diagnosis. I went in with very little knowledge and came out with a much better understanding of the

syndrome, and quite a bit of educational material. More than that, I came home knowing that I had a new family. One like no other, that would listen to me complain, help when I needed it, and bring a smile to my face like no other," said Holly. Aden is so lucky to have all of your boys to look up to, and I am so lucky to have all of you. There is more strength within this organization than I could have ever had alone. The knowledge you all have and the way you have put it to use has just been incredible. Thank you, for who you are, what you do, and how you do it. How do I feel overall?? Scared, nervous, anxious, but good that I am not alone." ~Holly Edsel

Condolences may be sent to:

Holly Edsel, 1232 Garren Creek Rd.,
Fairview, NC 28730

As for Laurie and Kevin Maxfield, it was though everything in their world came to a complete and sudden stop when they were told by their genetic counselor that Tanner, their five-year-old son, **might** have a serious medical condition called Barth syndrome. Immediately, the questions began, said Laurie. "What is the future for Tanner? How serious is this Barth syndrome? Is there treatment or even a cure? And the most difficult question of all, Can Tanner die from this syndrome?" The genetic counselor's response at this time was 'I



Kevin, Tanner and Laurie Maxfield

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...NEW HOPE AND A SENSE OF BELONGING

(Continued from page 17)

am not too familiar with this syndrome and I need to research more about it before we can answer a lot of the questions you have. I don't want to mislead you.'

Quickly swinging into survival mode, Laurie and her sister LeeAnn swept the Internet to learn more about this unfamiliar area. Everything Laurie read led her to believe that Tanner had no chance to live a normal life and that he might even die at an early age. "My life was consumed with the thought that my beautiful child could possibly be facing heart failure or other serious complications related to Barth syndrome," said Laurie. It was Laurie's sister who found BSF's website on the Internet, which literally changed their course of action. An email went out to Shelley Bowen, President of BSF and immediately Shelley responded with information of BSF's conference in Baltimore the following weekend. "We were told that other children with Barth syndrome would be there and that the leading scientists and physicians studying Barth syndrome would be in attendance."

"I have never experienced the emotion I felt in Baltimore with these wonderful people," said Laurie. "The feeling of true love, concern and commitment to these boys was overwhelming. Life didn't seem to be ending for these families. My hope grew with each boy I met, and I soon became aware of the fact that if Tanner did have Barth syndrome his life didn't have to end. I knew there was hope in the future."

Laurie and her family walked away from the conference with a great deal of knowledge about Barth syndrome. However, since returning home, they have been informed that their son Tanner's G4.5 test has come back with no genetic mutation. "This was a big sigh of relief," said Laurie, "...but in laymen terms we have been told that Tanner has a form of Barth syndrome, so the emotional roller coaster continues. The men and women of the Barth Syndrome Foundation are truly the heroes of today and hopefully I too can help make a difference, as you have made in mine." ~Laurie Maxfield

Veerle Van Langendonck and Erik Swennen are from Belgium and are the parents of two sweet little boys, Tuur (3) and Jef (1). Jef was born full-term but was a very tiny baby. At five days old, Jef's paediatrician heard an abnormal heart rhythm, and an echocardiogram was performed. Jef was immediately sent to an academic hospital, where he was diagnosed with serious hypertrophic cardiomyopathy of an unknown etiology. "A terrible period of uncertainty and "worst case scenario" had begun," said Veerle. Knowing that there had been many heart problems in Veerle's family history, a

Metabolic Specialist immediately thought of Barth syndrome being a possibility.

Many tests were conducted, but it was not until Jef was 6 months of age that he was officially diagnosed

with Barth syndrome. Veerle stated that, "On the one hand it was good that the 'unknown' was given a name, but on the other hand the words Barth syndrome represented such bad news because at that time, the only information that we knew was that boys died at a young age. From the first day of the diagnosis I have been more occupied with 'what can I do?' instead of 'why me?'" Veerle turned to the computer and started using the Internet for the first time. "Am I glad I did!" said Veerle. "I immediately found BSF's website and their announcement of the conference in Baltimore. That same night I was eagerly making plans for a trip to the USA!"

"Contact was also made with Dr. Peter Barth of the Netherlands, and genetic testing was done on my entire family and rather surprisingly one of my three brothers ([still living], as another younger brother died at four months of age) was also diagnosed with having Barth syndrome," said Veerle. "Paul is now 34 years old and has done rather well. We hang on to the hope that Jef will do as well as his Uncle Paul has done. My son Jef is definitely on the right track!! He is really just like his older brother Tuur, a cheerful active child, despite his cardiac problems. The only VISIBLE sign of his illness is his very small stature."

The contact with BSF has been a wonderful source of knowledge and support," said Veerle. "It was helpful to talk with someone 'on the same level.' It was so frustrating talking to friends or colleagues when I in fact myself could not explain properly what was going on or what was going to happen! Conversations always ended with: "but look, he is doing fine, everything will be OK..."

"I want to say 'I am so happy to be a part of this family,' said Veerle. "...but as BSF has done all the work, I feel like I don't deserve the word *part*; I rather feel *blessed* to have found these terrific people and I will do my best to help the foundation to fight for progress in medical research." ~Veerle Van Langendonck



Erik, Veerle, Tuur and Jef



THE MAKING OF A CONFERENCE

By Anna Dunn, Vice President and Family Liaison

“...It combined all the merits of the previous meeting in 2000 with a flawless and superb organization. I enjoyed the friendly and positive atmosphere between the families, between the scientists and between all of us together.”
~Peter G. Barth, MD, PhD

Eighteen months ago a discussion took place at a BSF Board meeting, in which the board members asked me if I would be interested in orchestrating the 2002 International Family and Scientific Conference. My immediate response was “yes,” for my heart and passion is with “our boys” and the BSF. All the while, my mind was saying, “what did you just sign yourself up for?” In working with the BSF I have acquired a few new talents, and coordinating the making of a conference was going to be my newest talent yet!

Orchestrating a conference involves many needs: lodging, meals/breaks, meeting space, audio visual needs, accommodating schedules, child care, social attractions, registration, clinics, consults, and the list goes on and on. To accomplish these tasks, I formed a Conference Committee of dedicated family members: Rosemary Baffa, Shelley Bowen, Liz

(Continued on page 24)

IMPORTANCE OF BSF'S REGISTRY

By Mark Dunn, Registry Coordinator

As many of you already know, The Barth Syndrome Foundation, Inc. (BSF) has created a Registry and has initiated the first steps towards populating it with information from each of our Barth families. In its simplest form, the Registry is nothing more than a database of information collected from our members. It consists of responses to survey questionnaires, which are geared towards specific topics germane to Barth Syndrome.

Of course, in reality it is much more than that! The BSF Registry will eventually become a large database of information, which we hope to collect from all of our members. It will serve as a powerful and useful tool to researchers, physicians and Barth families. The benefits of such a database will be numerous, as it will provide a centralized repository of detailed data, which can be accessed (upon approval of a formal request) by the scientists and researchers who require it in order to assist with their studies of Barth syndrome. These requests will be screened and will require approval by the BSF Board of Directors before access to the information is granted. The request must come from a recognized and reputable scientist, physician or research organization and must include some detail as to the specific project being researched and the intended use of the information provided.

In order for the Registry to be as effective as possible we need 100% participation. If each and every Barth family that is known to us responds faithfully to the survey questionnaires then the data will become much more meaningful to researchers and scientists who have a need to utilize it. Please bear in mind that the informa-

tion we obtain from this Registry program could potentially help to serve as the very foundation for the future treatment and cure of Barth syndrome!

We have already received tremendous praise and backing from our SMAB (Scientific and Medical Advisory Board) in regards to the potential benefits of this Registry. All of the survey questionnaires have been reviewed and approved by the SMAB and they are anxious to make use of the data that is collected. In fact, at the 2002 BSF International Family & Scientific Conference the group of cardiologists (headed by Dr. Barry Byrne) who participated in the cardiology clinics at Johns Hopkins were so impressed with the Cardiology portion of BSF's Registry questionnaires that they asked if all the families could fill them out and return that portion before the end of the Conference! So you can see that this Registry program has enormous potential.

It is our sincere hope that each and every Barth family will participate in the Registry and help us to reach our goal of 100% participation. We realize that the survey questionnaires can be lengthy and somewhat intimidating and require a good deal of time to complete accurately; however, we also strongly believe that in the long run the potential benefits of participation will certainly outweigh the initial time and energy required to complete the surveys. Please look for upcoming email notifications regarding the BSF Registry and how you can join and participate. If you should have any questions or comments regarding the BSF Registry please do not hesitate to contact Mark Dunn at: mdunn@barthsyndrome.org

~Mark Dunn



SUPPORT FOR MEMBERS JUST A 'SEND' BUTTON AWAY

By Lynda Sedefian, Editor

The Barth Syndrome Foundation (BSF) is continuously striving to improve the ways in which knowledgeable information and support is provided to those caring for one who has been diagnosed with Barth syndrome. Given the fact that our families reside in so many different parts of the world and that much of the correspondence that goes on between our families takes place over the Internet, President Shelley Bowen thought that perhaps BSF could offer our families an opportunity to partake in an interactive Internet-based discussion forum where specific questions and/or concerns could be addressed which pertain to the care and management of Barth syndrome.

With the assistance of the Genetic Alliance, such a tool has been created. In February of 2002, a "LISTSERV" was launched which allows for the internal communication and interaction between group members of BSF. This LISERV has allowed BSF a secure forum to facilitate ongoing discussions amongst our members, thus creating a unique opportunity for our families to access the knowledge of quality experts as it relates to the overall care and management of Barth syndrome.

This LISERV not only allows but also encourages interaction between members who are part of the group. Every member can write to the list, and in doing so, all members will receive a copy of the message. In this way, discussions can take place and views can be exchanged among a large number of people. To date, many different topics have been addressed which has stimulated much interaction among our group. BSF has been extremely fortunate to have a number of guests volunteer their time and expertise as it relates to their experience of Barth syndrome in an effort to provide a better understanding of this disorder. We must stress that although the level of expertise these guests offer is very comprehensive, please understand that this information does not constitute as medical advice.

BSF would again like to stress that this LISERV is a secure forum in that it is not open to the general public of the Internet. It is inclusive of only those families affected by Barth syndrome and professionals involved with this disorder. An invitation must be received by BSF's LISERV Manager Shelley Bowen in order for an individual to become a subscriber to the list. Once an invitation has been received, you will need to complete a form to subscribe to this LISERV. You will then be sent an email requesting confirmation, which will prevent others from gratuitously subscribing you. Once a member,

you may post a message to the LISERV at any time. All postings are then distributed to the entire group.

All messages posted to BSF'S LISERV are maintained in archives for the benefit of new subscribers. This also allows easy access to a previous discussion that you now may have an interest in. To access these archives you will be required to log into the mainframe of the LISERV. At that point you may retrieve information by either the "thread," "subject," "author" or "date." For example, suppose you are in the process of enrolling your son in an "Early Intervention" program and you are curious as to what types of services other children with Barth syndrome receive. At that point you may want to access the archives to search whether or not any previous discussions have taken place regarding this issue. In this case scenario, it might be best to research this topic of "Early Intervention" by "subject."

Listed herein are the varying topics that have been discussed thus far, along with the facilitator: *Overview of Barth Syndrome* (Peter Barth, MD, PhD); *Genetics of Barth* (Iris Gonzalez, PhD); *Occupational Therapy* (Raghad Schroeder); *Physical Therapy* (Ginny Torncello); *Educational Concerns* (Joan Stoner, EdD); *Emotional Well-Being* (Jules Spotts, PhD); *Phospholipids and Cardiolipin* (Grant Hatch, PhD/Michael Schlame, MD); *Cardiology*, (Barry Byrne, MD); *Metabolic Aspects of Barth Syndrome* (Richard Kelley, MD, PhD); *Pediatrician's Role* (Patricia Blanco, MD); *Siblings* (Alanna Layton/Jess Wilkins); *Isolation from Expert Care* (Jeanette Thorpe/Shelia Mann); *Making Time for Spouses* (Rosemary Baffa); *Becoming a Partner in the Management of Care* (Joke van Loo); *Coping With School* (Jason Downard/Sue Wilkins); *Living with Limits* (Rosemary Baffa); *Grandparents—An Involved Observer* (Moirra Masterson); *Dealing with Well-Wishers* (Shelley Bowen); *Being a Neurotic Mom* (Lynda Sedefian); *Becoming Pregnant after Having a Child with Barth Syndrome* (Michaela Damin); *Supporting Families in Your Region* (Carolina Reece/Anna Dunn); *Awareness in Your Community* (Steve Kugelmann); *Getting Involved with BSF* (Steve McCurdy); *Hobbies when there are Limits* (Maryellen Angeloni); *Family Secrets* (Rosemary Baffa); *Caring for a child with a heart transplant* (Darlene Guasco).

We thank those who have participated and encourage the continued internal communication and interaction, as it is through this type of communication that everyone involved benefits. December's topic will focus on Assistive Technology in Education (Joan Stoner/Eileen Juico).

~Lynda Sedefian



GRANDPARENTS' FORUM

By Dick and Sharon Olsen

The BSF 2002 Conference was very enlightening. We were most impressed by the organization of this conference, which consisted of an excellent panel of doctors who participated in both the scientific and family sessions. The panel discussions were excellent, and we were impressed by the panelists' dedication to Barth syndrome and straight-forward answers that were provided to families.

As a grandparent of a child with Barth syndrome, we had the privilege of being a part of the "Grandparent Forum" which stimulated frank and open discussions/concerns about Barth syndrome. The bonding with other grandparents helped us realize that we all share many of the same concerns. It was especially beneficial for the "first-time" grandparents, as they learned that they weren't alone and that there was hope.

Many issues were discussed which were of concern, such as feelings of guilt on the grandparent's part; experiencing double the pain, as we now experience pain not only for our child, but also for our grandchild. Knowing how strong and determined these parents are, we worry that they try to do too much, and we also worry about how living with a life-threatening disorder will affect the siblings of that child. Also of concern were problems that the children are experiencing in school, such as the lack of stamina that these boys experience and the fact that many are not able to attend a full day of school. The issue of working with the school to accommodate the "special needs" of a child, (Federal rules regarding handicapped children), and the decision to actually have your child "labeled" was also discussed.

Different ways in which we as grandparents could offer our assistance were discussed, such as being there emotionally for support, offering our time to watch the Barth child and siblings to allow parents one-on-one time, and assisting with the financial burden whenever possible.

Thank you for all your hard work, and we are looking forward to the next conference.

~Sharon Olsen



BSF IS BRANCHING OUT



The Barth Syndrome Foundation, Inc. (BSF), a USA-based not-for-profit organization established to support the needs of those individuals diagnosed with Barth syndrome and their families, is also committed to broadening outreach efforts worldwide. Those of us affected by Barth Syndrome know the hopes, fears, joys, and tears of this often fatal disease. We know that Barth Syndrome affects boys and their families everywhere; it is no respecter of boundaries, race or religion. With the world becoming smaller every day, we are truly an international community working closely toward our vision, "... that not one more child will suffer or perish from Barth Syndrome."

At BSF's 2002 conference, which proved to be memorable as well as inspirational to those in attendance, a seed was planted to expand the growth of BSF for the following reasons: (1) to broaden our ability to increase awareness; (2) to provide local support and country-specific information to affected families; and (3) to establish additional BSF affiliates as charity organizations who will be able to raise much-needed funds to further research efforts.

"The energy of the dedicated BSF members gave way to the opportunity of BSF Board members meeting with our group of Canadians to discuss the desire to develop a BSF "Trust" in Canada," said Catherine Ritter, who will hold the position of "President" of the BSF Canadian 'Trust.' "With the help and guidance of our American counterparts we will develop our mandate, goals and direction. The preliminary work is already underway! A lawyer has kindly offered to donate his time in guiding us through the legalities of incorporating and acquiring charitable organization status. We look forward to the challenges that are ahead and I encourage all those affected to join and support BSF as we strive for awareness, knowledge, and a cure."

~Catherine Ritter—Ontario, Canada

"It is also with great pleasure that we can now announce the process of starting a BSF affiliated charity group in the United Kingdom," stated Michaela Damin, who will hold the position of President for BSF's affiliate in the UK. "We are very lucky to have the help of an attorney here in the UK who is setting up the Trust Deed for us. He has kindly provided his services free of charge and is currently putting the finishing touches on all official documentation. Another exciting announcement is that we will have the active participation of Dr. Colin Steward from the Bristol Royal Hospital for Sick Children, who has extensive experience with regard to diagnosing Barth syndrome. Dr. Steward is very committed to raising awareness about this often under-diagnosed disorder. Our top priority at present is to identify affected families around the world and to pool our resources together. We welcome offers of assistance, advice and cooperation from any interested party. Every day, we move one step closer to our vision, "a world in which not one more child will suffer or perish from Barth Syndrome."

~Michaela Damin—Wiltshire, England



OVER \$289,218 RAISED IN 2002, WITH MORE TO COME!

By Steve McCurdy, Vice President, Finance & Development

What a year for fund raising, and its not over yet! With the help of a growing list of committed volunteers and contributors, BSF has raised over \$289,218 for research, awareness and family support through October. To place this feat in perspective, we initially set ourselves the ambitious goal of raising \$785,000 over a five-year period. In less than two years, we have already raised almost \$500,000, 64% of our five-year goal! The generosity of our friends leaves me breathless... and thankful!

Our volunteers have used walk-a-thons, marathons and triathlons; golf tournaments and car rallies; raffles, auctions, house parties and solicitation letters. Contributions have come from families, friends, colleagues at work, friends of friends, businesses and foundations in over 25 states and 5 countries. We've received contributions ranging from \$4 to \$100,000. They have come by check, cash and credit card, delivered in person, by mail and via the Internet. Over 444 individuals and institutions have contributed their money to BSF so far in 2002. Over 100 more have made contributions "in-kind", offering their products and services, their time and expertise to help us achieve our goals. You can see an impressive list of names at the back of this newsletter – so many that they cover two pages!

Most people find asking other people for money to be an embarrassing task to be avoided at all costs. But when the cause is as important as increasing awareness and finding a cure for Barth syndrome as it is to all of us, and when the volunteers find such fun and inspiring ways to ask, then a potentially distasteful task turns into a rewarding and positive experience for everyone. The brief stories that follow (in addition to the letter-writing appeals and marathons which I have described previously) cannot do justice to the contributions made by our intrepid fundraisers. Their collective efforts have enabled us to accelerate all of our programs, including research, and have enhanced the credibility of BSF with institutions like the NIH and other groups critical to our future success. We owe these people and their friends our most sincere thanks and appreciation... and continued success!

House Parties and Foundation Grants

Last Christmas, Rosemary Baffa opened her house to family and friends for the holidays to benefit BSF and raised several thousand dollars in donations. The Baffas have also been instrumental in helping BSF find and convince much larger foundations to take an interest in BSF and contribute significant grants. You may remember

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	<u>Year To Date October 31</u>	
	<u>2002</u>	<u>2001</u>
STATEMENT OF REVENUE AND EXPENSES		
Donations	\$ 289,218	\$ 137,432
Other Income	\$ 3,587	\$ 1,317
Total Revenue	\$ 292,805	\$ 138,749
Administration	\$ 12,420	\$ 4,892
Family Support & Education	\$ 13,567	\$ —
Fundraising	\$ 6,422	\$ 484
Physician & Public Awareness	\$ 12,651	\$ 1,554
Research	\$ 2,347	\$ —
Total Expenses	\$ 47,407	\$ 6,930
Excess or (Deficiency) of Revenue Over Expenses	\$ 245,398	\$ 131,819
	<u>Year To Date October 31</u>	
	<u>2002</u>	<u>2001</u>
BALANCE SHEET		
Total Assets	\$ 386,247	\$ 132,516
Unrestricted Funds	\$ 284,907	\$ 32,516
Restricted Funds—Research	\$ 100,000	\$ 100,000
Restricted Funds—Conference	\$ 1,340	\$ —
Total Liabilities and Fund Balance	\$ 386,247	\$ 132,516

OVER \$289,218 RAISED IN 2002, WITH MORE TO COME!

(Continued from page 22)

that we received an anonymous grant of \$100,000 last year from a major foundation. The same foundation has been so impressed with our progress, particularly with our Scientific and Medical Advisory Board and our approach to research grants, that they gave us a second \$100,000 grant again this year! Their vote of confidence gives us enormous credibility both when we approach other foundations and with the research community. Thank you, Rosemary and Mary Baffa!

Lynda and Derek Sedefian "Walk" with Their Friends for the Second Year



Voorheesville Walks to Raise Awareness

For the second year, Lynda and Derek Sedefian and their many supporters in Voorheesville, NY held their April Barth Syndrome Walk-a-thon, raising over \$14,500 and increasing awareness of

Barth syndrome. The walk-a-thon has become a wonderful annual event for the members of this small community, as well as the Albany County Sheriff's Department who accompany the walkers. Once again, Lynda was supported by the local Kiwanis Club and VFW chapter as well as Wal-Mart, Sam's Club and several other businesses. Lynda sold raffle tickets and held an auction to add to the donations from the walkers. And she has put together a wonderful "how to" guide available to any others who would like to host a similar event. Don't be shy!

First Annual Barth Syndrome Golf Outing

Jan and Steve Kugelmann organized a charity golf outing in their hometown of Merritt Island. 144 golfers paid \$55 each to play a round of 18 holes and enjoy a lunch sponsored by a local restaurant. Local companies sponsored individual holes, golfers bought



Michael Bowen (L) and R.J. Kugelmann (R) - birdie or bogey?

"mulligans" or free shots, Jan and Steve held an auction and ran a raffle and many of the participants made extra donations. I should add that Steve was very effective in getting his employer to help him raise funds for BSF and to increase awareness among employees. Shelley and Michael Bowen, native Floridians themselves, also attended. Everyone had a great time, the Kugelmann's raised almost \$20,000 for BSF and Merritt Island is now much better informed about Barth syndrome. All in all a fine afternoon!

Car Rally



Sheila and Benjamin Mann are presented with a check from Sam's Club

How many of us are a part of a local club centered on a favorite hobby? David and Shelia asked David's car club to help them raise funds for BSF, and the club was only too glad to help.

They already knew that the Mann's son Benjamin had Barth syndrome, and their annual car rally was often used to raise funds for charity. BSF was a perfect fit. Shelia also held a raffle to raise additional contributions. Their efforts raised \$3,000 for BSF, and heightened Barth awareness in Chattanooga, TN.

Ironmen Race for Barth Syndrome

Gary Rodbell and John Steigerwald are a couple of unusual fellows. They like to swim, bike and run. Over long distances. Very long distances! Like 2.4 miles (swim), 112 miles (bike) and 26.2 miles (run) back to back on a single day. They are Ironmen. But more to the point, on October 9th in Panama City, Florida, they were our Ironmen! And on that day, not only did they com-



The Janus Charity Challenge is a program that is designed to encourage athletes to use the Ironman qualifying events as a platform for raising financial support for their favorite nonprofit organization.

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OVER \$289,218 RAISED IN 2002

(Continued from page 23)

plete the full course of over 140 miles in 16 hours and 10 minutes and 14 hours and 36 minutes, respectively, but they also raised \$74,000 in donations, and won an additional



From left to right: Gary Rodbell, Will McCurdy and John Steigerwald

\$5,000 from the Janus Funds Charity Challenge – a total of \$79,000 for the Barth Syndrome Foundation. Our Ironmen turned their strength and endurance to gold for the BSF. A portion of Gary's letter requesting financial support for the race explains his reasons best:

"A very good friend of mine is 16 years old. I've known him and his family since he was born. Unfortunately, he was born with a very rare genetic disorder called Barth syndrome, which weakens his heart, his skeletal muscles and his immune system. He is one of the oldest surviving boys with this disorder in the world. He can't participate in sports, has enough endurance to attend only one or two periods of high school each day, and spends way too much time with doctors for a kid his age. But I think he may be stronger



Will & Michael extend a high-five to Gary while passing by

than I am. He has to put up with fatigue and pain every day but he doesn't have a race to look forward to, to justify putting his body through hell. I choose to run the risk of a sprain or a strain... he lives with the risk of heart failure or infection without having a choice. Yet every time I see him, all I see is a smile and an undaunted spirit that I truly envy. He

understands what it means to not give up. To keep going no matter how lousy you feel. And so I am training and running this race for him."

Will McCurdy, Michael Bowen and their Dads were there to cheer them on, while Shelley Bowen delivered updates to the Barth families over the ListServ. Families from Australia to Canada tracked the progress of our Ironmen throughout their grueling race. We are lucky to have a couple of Ironmen like Gary and John racing to benefit our boys who must sometimes themselves be "Ironboys" as well. I guess these guys all stick together!

~Steve McCurdy



MAKING OF A CONFERENCE

(Continued from page 19)

Higgins, Jan Kugelmann, Kate McCurdy, Shelia Mann, and Lynda Sedefian. They were my right and my left hand, and they inspired me to work even harder for "our boys." It was Rosemary, Liz, Jan, Sheila, and Lynda who focused on gathering in-kind donations; contracting for audio visual needs as well as obtaining supplies that were included in "Emergency Care Bags"; gathering educational information which was provided at the "Activity Fair" and scheduling attractions for the families while in Baltimore; and coordinating BSF "Saturday Night Social", respectively. A special thanks to the grandmothers on board who used their talents in making this conference one of giving the gift of love. Joyce Lochner, Derek Sedefian's grandma, handmade "wall hangings" which included the names of our "Barth boys/young men." These were presented to Drs. Barth and Kelley on behalf of the families as our gift of appreciation. Sharon Olsen, R.J. Kugelmann's grandma, created the beautiful canvas bags with BSF's logo, which are now known to us parents' as our "Emergency Care Bag."

"I admit, that in the past I had some reservations about the value of having scientific and family meetings at the same time and place, but this meeting has convinced me that this formal and informal exchange is extremely fruitful."

~Peter G. Barth, M.D., Ph.D.

The BSF 2002 Conference was a huge success on many levels. In attendance were 100+ Barth family members present from Canada, Belgium and the U.S. The Scientific and Family Sessions encompassed the participation of approximately twenty-seven scientific/educational participants combining clinical cardiology, hematology, and neurology with basic genetics, biochemistry, mitochondrial bioenergetics and gene therapy.

Cardiology, Math Skills Development, and Neurology clinics (Research Studies) were provided to all families, as well as Cardiology, Neurology, Metabolics; He-

matology; Genetics; Education; and Nutrition consults. The information gained from these clinics/consults is invaluable not only to BSF, but to future research. In hindsight, I am left with an enormous sense of awe and thankfulness. I feel privileged and honored to be a part of BSF, and to come to know and work with so many wonderful Barth families who share the passion with me for "our boys". Thank you for the opportunity to reach new horizons. I know you share my wishes in hoping for the day when we can confidently say that "...not one more child will suffer or perish from this disorder."

~Anna Dunn



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A SIBLING'S UNDERSTANDING OF ...

By Alanna Layton—Sister of Michael Bowen

Let me start off by saying that I had a blast at the conference. I thought everything went very well and it was obvious that everyone had worked so hard to make it a success. It was wonderful to see everyone again and to meet new people. The biggest thrill was to see all of the Barth boys together. I will never fail to be amazed whenever I see all of them together, no matter how many times it may be. To see so many boys who have Barth syndrome in one setting, when years ago we knew of no other living person with this disorder, is truly awe inspiring.

When I first arrived at the conference, I felt a bit awkward. Being an adult sibling I thought, "Okay, I'm not a child anymore so I can't be with the children yet I don't really belong in the parent's sessions." I didn't know if I could really contribute anything, so I felt relieved when I was asked to sit at the desk. I never expected to be taking over childcare of some of the children, though, but I enjoyed having them around!

What impressed me the most about the conference was observing the level of maturity and sensitivity among the siblings who were there. It was especially interesting to see the differences among older and younger sisters. Older sisters tended to be more maternal and protective of their brothers, while younger sisters seemed to have the attitude, "So you're sick. Get over yourself!" The one constant factor among all the siblings was their kindness and affection towards their Barth brothers. It really touched me to watch one older sibling rocking her brother and humming to him when he was upset or to witness another hug her brother and comfort him when he cried.

I may be older than the majority of the siblings but it was cathartic for me to be around them. It is so comforting to know that these siblings don't have to grow up feeling isolated. They have other Barth siblings they can relate to and, thanks to e-mail, communicate with on a regular basis if they choose. I grew up without the privilege of knowing a Barth



Alanna and Michael share a special moment!

sibling. My parents went through "diagnosis purgatory", as Mom called it, for 30 months before we received an accurate diagnosis. It was difficult for me to watch my brothers being admitted to the hospital on a regular basis and to not have them around as much as I wanted. I know I could have used a friend who understood the tumult occurring in my life at that point. Now that I am grown, I feel like it is important for me to be a source of encouragement and support to these siblings if they need it. In that way, my experiences will not have been in vain.
~Alanna Layton

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