Barth Syndrome Foundation

NEWSLETTER

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BARTH SYNDROME FOUNDATION AWARDS FIVE RESEARCH GRANTS

By Kate McCurdy, Vice President, Science and Medicine

It is my great pleasure to announce that the Barth Syndrome Foundation, Inc. (BSF) has awarded five research grants from our first cycle of applications. Our financial commitment to these projects, which will be conducted over the next one to two years, totals $148,942. We are extremely fortunate to have a number of top-notch laboratories around the world interested in conducting research specifically on Barth syndrome, and we on the Board of Directors of BSF are very gratified that we will be directly supporting five of these. We now are sponsoring studies focusing on various aspects on Barth syndrome in three countries: The Netherlands, Canada and the United States. The work that will be done in these laboratories by both Ph.D.s and M.D.s has a broad range. Most of the projects relate in some way to the quest for further insight into the molecular mechanisms that underlie Barth syndrome. In other words, they will focus on such questions as: What happens at a molecular level inside the cells of a Barth patient as the result of a *TAZ1* (or *G4.5*) gene mutation? What exactly is the function of the *TAZ1* (or *G4.5*) gene and what does it do differently (or not at all) in a Barth patient's body? How does the biochemistry of all this work? Are there proteins that should be produced that are not or are different products made? If so, how do they fit into other biochemical or genetic activities of the body? Does this gene affect other genes? In addition, a final project has to do with trying to create a mouse model of Barth syndrome so that studies can be conducted on *mice* with Barth syndrome before they are confirmed in *humans*. Though very similar to humans genetically, there is no guarantee that the mouse will survive once it has been created. But we have one of the best labs in the world (that has a great deal of similar work supported by the National Institutes of Health) working on it, so that work is in very good hands.

In addition, we also are exceedingly pleased that Michael Schlame, M.D. had a large grant application accepted by the American Heart Association just as we were reviewing the grants submitted to BSF (including his). As a result, he withdrew his BSF application entitled *Biochemical Basis of Barth Syndrome*, since it was for just a portion of the work that now has been fully funded by the AHA. We congratulate Dr. Schlame on his success with this! Please look on page 7 of this newsletter for an article about his work.

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2002 Year-End Report

By Valerie ("Shelley") Bowen, President
Barth Syndrome Foundation, Inc.

2002 marked our second complete year since the Barth Syndrome Foundatuion was incorporated. For a relatively young foundation, I am proud to be able to report that we have made great progress against every one of our program goals. This could never have happened without the dedicated support of our growing list of volunteers, scientific and medical advisors and contributors. The Officers and Directors of your foundation and all of the families who are affected by this disorder remain eternally grateful to all of you who have contributed so much of your time and resources to help us achieve our goals - ***"…to guide the search for a cure, to educate and support physicians and to create a caring community for affected families."***

Shelley Bowen

What follows is a summary of our accomplishments in 2002, organized under our major program goals.

Program Development

1. **To insure that all appropriate physicians are aware of Barth syndrome, have ready access to the latest information to insure an accurate diagnosis and can easily make use of the medical resources they need to deliver successful treatment.**
   a. Launched awareness campaign initial results:
      i. Associated Press (AP) article describing Barth syndrome and featuring Lynda and Derek Sedefian was published in over 50 newspapers nationwide
      ii. Six additional articles were published in local papers around the US, each featuring a local family affected by Barth syndrome. Many of these articles also reported on local BSF fund raising activities
   b. Hosted BSF International Scientific Conference with over 60 physicians and scientists present, more than doubling the scientific attendance at the first conference
   c. Increased visitors to the BSF website. Total volume of visits in 2002 rose to 28,685. Total number of new (unique) visitors in 2002 increased to 6,880
   d. Published two BSF Newsletters in 2002 including a December issue featuring an article by Dr. Colin Steward, Ph.D. entitled "*Barth Syndrome…a Much More Common Diagnosis than Generally Recognized"*
   e. Presented a parent's perspective of Barth syndrome to Sarah Lawrence College for genetic counseling second year graduate students and University of Chicago school for genetic counselors
   f. Presented a parent's perspective of Barth syndrome to over 200 physicians at AMC (Academic Medical Center), in Amsterdam
   g. Developed relationships with the Office of Rare Diseases (ORD), a division of The National Institutes of Health (NIH), and the National Institute of Neurological Disorders and Strokes (NINDS), also a division of NIH

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h. Initiated *Barth Syndrome Registry* to collect family and medical data about Barth syndrome and those it affects. This is the only comprehensive, longitudinal database on Barth syndrome in the world and will be an invaluable resource for researchers and clinicians

i. Created a world-class BSF Scientific Medical Advisory Board including (13) scientists and physicians and (1) ex officio BSF Board member

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

a. Co-sponsored BTHS Cardiomyopathy Research Study with Dr. Barry Byrne, Dr. Carolyn, Spencer, Dr. Reid Thompson and Dr. Philip Spevak

b. Launched BSF Research Grant program (with our RFP being distributed to at least 2,200 researchers and institutions internationally) and awarded (5) grants with a funded total of $109,312 for BTHS research

c. Continued collaboration with Michele Mazzocco Ph.D. at Kennedy Krieger Institute at Johns Hopkins University, for research in the Math Skills Development Project including boys with Barth syndrome

d. Initiated the development of a set of management guidelines in the medical care of a child with Barth syndrome in cooperation with a team of scientists and clinicians

3. To create a caring community that will offer each Barth Family information, guidance and emotional support

a. Hosted BSF Family Conference in Baltimore including clinics, lectures and panel discussions featuring world experts in Barth syndrome with over 100 family members in attendance

b. Created the BSF Listserv serving 114 interested families, clinicians and scientists, and featuring 33 topic leaders (including physicians, educators, parents, genetic counselors and scientists) in 2002

c. Published two issues of the BSF Newsletter featuring information of great use to families affected by Barth syndrome

d. Increased BSF family membership by 34%

e. Awarded (2) computers to BSF families in need to insure that they can enjoy the full benefits of BSF delivered via the internet

4. To build and sustain a broad base of concerned contributors who will provide the funds needed to accomplish our Mission and Goals

a. Raised in excess of $328,000, a 125% increase over last year, from over 500 contributors, also an increase of 66% over 2001

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2002 Year-End Report

b. Received a $100,000 donation from an anonymous, dedicated friend

c. Arranged to accept all major credit cards insuring that donations can be accepted from around the world, in any currency

d. Obtained funding for BSF Family Conference Sessions from The Lied Foundation Trust, Christina M. Hixson, Trustee; and for the Scientific Sessions from the Office of Rare Diseases (ORD), a division of The National Institutes of Health (NIH) and the National Institute of Neurological Disorders and Strokes (NINDS), also a division of NIH

5. To create, inspire and make effective use of an organization of volunteers dedicated to reaching our vision.

a. Increased distribution of BSF Newsletter from 500 to 1,300 in one year

b. Hosted the first BSF volunteer workshop inviting 20 highly motivated volunteers to help plan and delegate responsibility for the execution of the BSF Program Goals

c. Welcomed Michael Wilkins as an officer and new Treasurer of BSF, Inc.

d. Initiated development of two International Affiliates of BSF, Inc. to be called:

i. The Barth Syndrome Trust, United Kingdom *(Michaela Damin, President)*

ii. The Barth Syndrome Foundation, Canada *(Cathy Ritter, President)*

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...BSF Awards Five Research Grants

You can see that BSF has had a fabulous inaugural year of its grant program. I want to offer my sincere thanks to those who submitted applications and to those on our Scientific and Medical Advisory Board (SMAB) and those outside BSF who helped with this process. I also want to thank each and every individual who sponsored a fundraising activity or who gave BSF financial support, because without the funds you have helped us gather, this very important scientific work would not be proceeding. We look forward to learning what knowledge is gained from these projects and what progress toward the understanding of Barth syndrome, its treatment and ultimately a cure can be made as a result.

The five proposals that have been accepted by BSF are summarized as follows:

Principal Investigator: Iris L. Gonzalez, Ph.D.; Senior Research Scientist
Institution: A.I. DuPont Hospital for Children; Wilmington, DE
Amount: $13,310
Time Period: 1 1/2 years
Title of Project: *A Study of TAZ mRNAs in Barth Syndrome Individuals*

Abstract:

"The TAZ gene is complex in the sense that its primary transcript (pre-mRNA) undergoes multiple alternative splices and can potentially give rise to different-but-related proteins. Additional complexity comes from its pattern of expression: not all of the alternatively-spliced mRNAs are found in all tissues. We first plan to characterize all the mRNA variants present in normal white blood cells, where the most variants were described by the discoverers of the TAZ gene; following this, we plan to examine and compare the mRNAs of Barth syndrome individuals with mutations in different parts of the TAZ gene to see which forms are present or absent. We can then ask questions that could lead to a better understanding of the syndrome, such as 'Does a specific deletion/frame-shift mutation lead to loss of all TAZ mRNAs or do we find alternatively-spliced mRNAs that skip the affected exon?'

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BSF Awards Five Research Grants

To accomplish this aim, we will obtain mRNA from normal and from Barth syndrome individuals. This RNA is a mixture of all products characteristic of the chosen tissue and we will perform TAZ-specific amplification and cloning (expecting to obtain cloned representatives of all the kinds of TAZ mRNAs that are present) and perform DNA sequencing to determine which exons are present/absent. The results from Barth syndrome individuals will be compared to each other and to those of the normal controls.

The results will show which TAZ gene products are present in the subjects' blood cells and which tafazzin proteins may potentially be made in these cells. It is likely that, depending on the mutation, some of the TAZ products are still made and are normal. Eventually, such data may lead to more accurate correlations of specific gene defects with disease severity. Correlation with the protein, biochemical and functional studies carried out by others should also be possible."

Principal Investigator: Miriam L. Greenberg, Ph.D.; Professor, Biological Sciences
Institution: Wayne State University; Detroit, MI
Amount: $39,630
Time Period: 2 years
Title of Project: *TAZ1 Gene Function in Yeast: a Molecular Model for Barth Syndrome*

Abstract:

"Barth syndrome is a genetic condition associated with a mutation in the gene G4.5. Recent studies indicate that the mutation in Barth syndrome leads to a defective remodeling of cardiolipin (CL), a phospholipid that is required for optimal mitochondrial function. How can the defective remodeling of CL lead to aberrant cell function? The yeast model provides us with a powerful tool with which to address this question. The yeast *taz1* mutant has a mutation in the yeast gene that is homologous to the human G4.5 gene, and like Barth syndrome cells, cannot properly remodel CL with unsaturated fatty acids. We propose to identify aberrant physiological conditions that stem from the mutation in *taz1*. Our specific aims are as follows: 1) Determine the role of *TAZ1* in mitochondrial functions known to require CL; 2) Use a genetic approach to identify pathways that require a functional *TAZ1* gene; and 3) Determine the role of *TAZ1* in CL biosynthesis.

Yeast is the most tractable organism in which to carry out this study, because it is more amenable than any other eukaryote to a combination of genetic, molecular, and biochemical experimental approaches. Furthermore, even mutants with defective mitochondria and the consequent inability to respire can grow if supplied a fermentable carbon source. Finally, the genes for CL synthesis in yeast have been well-characterized, and mutants are available. Because conservation of function has been demonstrated from yeast to humans in essential cellular processes, we expect that the outcome of these experiments will contribute significantly to our understanding of Barth syndrome."

Principal Investigator: Grant M. Hatch, Ph.D.; Professor, Pharmacology & Therapeutics
Institution: University of Manitoba; Winnipeg, Manitoba, Canada
Amount: $18,287
Time Period: 1 year
Title of Project: *The Molecular Mechanism of Barth Syndrome*

Abstract:

"In patients with Barth syndrome, the ability to remodel cardiolipin (CL) is reduced leading to a reduction in CL levels. Since acyltransferases are enzymes that remodel phospholipids and the Barth syndrome gene G4.5 codes for an acyltransferase, we hypothesize that a reduced ability to remodel CL could be one of or the underlying molecular mechanism responsible for Barth syndrome. The overall objective of the proposed research outlined in this application is to characterize the regulation of CL remodeling in COS cells transfected with a Flag-tagged cDNA coding for tafazzin acyltransferase(s) and in Epstein-Barr

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virus transformed lymphoblasts from Barth syndrome patients. The specific objectives of the proposed research are to: 1) Characterize the tafazzin acyltransferase(s) in COS cells transfected with the cDNA coding for tafazzin -- The subcellular localization of the tafazzin acyltransferase(s) will be determined by examination of the presence of the Flag-tagged protein in subcellular fractions prepared by classical subcellular fractionation techniques and western blot analysis using anti-Flag antibody. In addition, immunoflourescent confocal microscopy will be used to confirm the presence of the tafazzin acyltransferase(s) in that fraction. The kinetic parameters of the tafazzin acyltransferase(s) in that fraction will then be determined. 2) Characterize the tafazzin acyltransferase(s) in lymphoblasts obtained from Barth syndrome patients -- From the characterization studies in COS cells we will have developed the appropriate conditions for characterization of the tafazzin acyltransferase(s) in Epstein-Barr virus transformed Barth lymphoblasts. We anticipate that tafazzin acyltransferase(s) activity will be reduced in these lymphoblasts compared to controls. We will confirm that the reduction in tafazzin acyltransferase(s) activity corresponds with a reduction in CL remodeling using pulse labeling of cells with radiolabeled fatty acid precursors.

From these proposed experiments we will learn: 1) In what subcellular fraction the tafazzin acyltransferase(s) resides and 2) if the activity of the tafazzin acyltransferase(s) is reduced in transformed Barth syndrome lymphoblasts."

Principal Investigator: Arnold W. Strauss, M.D., Professor and Chairman of Pediatrics
Institution: Vanderbilt University
Amount: $40,000
Time Period: 1 year
Title of Project: ***A Mouse Gene Ablation Model of Barth Syndrome***

Abstract:

"Barth syndrome (MIM 302060) is an X-linked disorder that in its complete spectrum of manifestations is characterized by infantile-onset dilated cardiomyopathy, cyclic neutropenia, skeletal myopathy, diminished linear growth, and 3-methyl-glutaconic aciduria. Genetic analysis linked Barth syndrome to the tafazzin or G4.5 gene on chromosome Xq28. Human mutations in this G4.5 gene cause Barth syndrome, and more recent studies prove that mutations are present in individuals who do not exhibit the full spectrum of the disorder. That is, some males with only dilated cardiomyopathy, but with normal neutrophil counts and lacking 3-methyl-glutaconic aciduria, also exhibit G4.5 mutations. Thus, G4.5 mutations may be a much more common cause of pediatric cardiomyopathy than previously suspected. The pathogenetics or mechanisms by which G4.5 mutations cause the clinical manifestations of Barth syndrome remain unclear. Moreover, the basis for variation in clinical phenotype is uncertain.

The function of the G4.5 gene is also unknown, although homology analyses reveal that it is related to fatty acyl-transferases. Recent results show dramatic reduction in cardiolipin, a mitochondrial membrane phospholipid, in Barth syndrome patients' fibroblasts, consistent with the G4.5 gene being critical in phospholipid remodeling. These studies suggest that mitochondrial structure and function are abnormal in this disorder, especially in highly oxidative tissues such as heart and skeletal muscle. Because Barth syndrome is relatively rare, clinical investigations into the mechanisms underlying the disease have been limited in scope. No animal model of Barth syndrome exists. The clinical manifestations of Barth syndrome, with the exception of neutropenia, are also common in disorders of mitochondrial energy production, including those caused by mitochondrial DNA mutations, respiratory chain gene mutations, and disorders of fatty acid oxidation.

Because the major clinical phenotypes involve sarcomeric tissues and because mitochondria are abnormal, we propose the *hypothesis that mutations in the G4.5 gene alter energy metabolism by interfering with mitochondrial inner membrane phospholipid and cardiolipin composition, reducing ATP production from long chain fatty acids through alterations in mitochondrial respiration.* To examine various pathogenetic mechanisms, we propose the following specific aims:

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Specific Aim 1: To create a peri-natal tafazzin gene ablation using *cre*-inducible methodolgy.

Because a closely related homolog of human G4.5 is present on the mouse X-chromosome, it is likely that this protein performs similar functions in mice. Thus, we can elucidate G4.5 function by ablating this gene using *cre*-recombinase and loxP methodology that will allow normal synthesis of the X-linked gene during *in vitro* recombination and after blastocyte injection during *in utero* life. Given our particular interest in cardiomyopathy, we will generate *cre*-recombinase inducible global and cardiac-specific inactivations of the tafazzin gene.

Specific Aim 2: Analysis of tafazzin knockout phenotypes.

We will examine life span, exercise capacity, cardiac rhythm, myocardial function, neutrophil counts, and phospholipid composition in tafazzin knockouts of mice consuming a normal laboratory diet and physiological characterization with a mouse treadmill, serial echocardiography, telemetry with and without exercise, stress of exposure to cold and fasting, complete blood counts, and analysis of tissue and mitochondrial phospholipid compositions and respiration."

Principal Investigator: Frederic Vaz, Ph.D., Laboratory of Genetic Metabolic Disorders
Institution: Academic Medical Center; Amsterdam, The Netherlands
Amount: $37,715
Time Period: 1 year
Title of Project: *Resolution of the Function of the TAZ-gene and Characterization of its Gene Products*

Abstract:

"X-linked cardioskeletal myopathy and neutropenia (Barth syndrome, BTHS, MIM#302060) is an X-linked recessive disorder, which is characterized by infantile or childhood onset of dilated cardiomyopathy, skeletal myopathy, short stature and intermittent neutropenia. The gene mutated in this disorder is the *G4.5* gene of tafazzin (TAZ) gene, which is localized on Xq28. This gene shares homology with a family of acyltransferases, which are involved in phospholipid biosynthesis and/or remodeling, however, the exact function of the TAZ gene remains to be elucidated.

It recently has been shown that BTHS patients suffer from a cardiolipin deficit although their biosynthetic capacity to synthesize this lipid from its precursor phosphatidyglycerol is entirely normal. The defect in BTHS appears to occur after cardiolipin synthesis, when the acyl-chains are remodeled by a deacylation-reacylation cycle to obtain the correct cardiolipin species. It is this remodeling process which appears to be disturbed in BTHS.

The first aim of this research proposal is to elucidate the function of the TAZ gene and investigate the role of the different splice variants. To this end, we intend to clone the various TAZ cDNAs, express them in a heterologous expression system, and subsequently determine whether the different protein products exhibit acyltransferase activity toward monolysocardiolipin. Furthermore, we will investigate the levels of the splice variants in different tissues, both on the mRNA as on the protein level. To allow detection of the protein product(s) of the TAZ gene, an antibody is being raised against recombinant tafazzin. The second aim of this research proposal is to resolve the role of cardiolipin in oxidative phosphorylation and to investigate the recently discovered beneficial effect of linoleic acid supplementation. To achieve this, fibroblasts will be incubated with or without supplemental linoleic acid and the oxidative capacity will be determined using a variety of different techniques."

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

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

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

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

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

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

American Heart Association Awards Grant in Support of Barth Syndrome Research

By Michael Schlame, M.D., Dept. of Anesthesiology
NYU School of Medicine

A few months ago our laboratory received a grant from the American Heart Association to investigate the molecular mechanism of Barth syndrome. Since the first documentation of the disease by Peter Barth in 1983, many years have passed without much progress in its molecular pathophysiology. One significant discovery was made in 1996, when Daniela Toniolo's group from Pavia, Italy identified the X-chromosomal gene that is mutated in children with Barth syndrome. One year later Andrew Neuwald, working with National Institutes of Health in Bethesda suggested that the "Barth" gene belonged to a family of genes involved in lipid metabolism. Finally, in 2000 Peter Vreken and his co-workers at Emma Children's Hospital in Amsterdam showed that the affected lipid was cardiolipin, a specific substance in the powerhouses of cells.

The grant given by the American Heart Association is supporting research that builds on the discoveries made by Toniolo, Neuwald, and Vreken. We want to find out which lipids are affected in Barth syndrome. Is it only cardiolipin, or is the disease "spread" among other lipids? Furthermore, we want to find the missing link between the gene defect and the lipid defect. Children with Barth syndrome have lipids with the wrong fatty acid composition. Thus, we hypothesize that the missing link may be a protein that transfers fatty acids from one lipid to another. Finally, we want to understand how the fatty acid composition of lipids is related to the characteristic symptoms of Barth syndrome. This is probably the most ambitious part of the project because very little is known about the physiologic effect of abnormal lipids.

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2003 REQUEST FOR RESEARCH PROPOSALS

The Barth Syndrome Foundation, Inc. (BSF) is pleased to announce the availability of funding for research on the natural history, biochemical basis, and treatment of Barth syndrome.

Background

Barth syndrome is a serious X-linked recessive condition associated with cardiomyopathy, neutropenia, skeletal muscle weakness, exercise intolerance, growth retardation, and diverse biochemical abnormalities (including defects in mitochondrial metabolism and phospholipid biosynthesis). Because many clinical and biochemical abnormalities of Barth syndrome remain poorly understood, we are seeking proposals for research that may shed light on any aspect of the syndrome. We are determined to find improved treatments – and ultimately a cure – for this rare and underdiagnosed disorder.

Types of Proposals Sought

We are most interested in providing "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 are especially interested in attracting new investigators to the very interesting field of Barth syndrome research.

Funding

We anticipate awarding up to $150,000 in 2003, divided among several one- or two-year grants of up to $40,000 each. Funds will be available in December 2003, as soon as the successful grant applicants have been notified.

Process

We have a simple two-stage grant review process. First, letters of intent are submitted to the BSF Scientific and Medical Advisory Board for review. The letters of intent must include a completed information form, a clear and succinct two-page summary of the proposed research, and biographical information about the principal investigator(s). Then, based on the recommendations of the Scientific and Medical Advisory Board, the BSF Board of Directors will invite selected investigators to submit full grant applications of 10 to 15 pages, due on October 1, 2003. Please consult our website, **www.barthsyndrome.org** for further guidelines and application details as well as a listing of grants that BSF has awarded to date.

Deadline

The deadline for submission of letters of intent from interested investigators is **June 27, 2003**.

Contact Information

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Cardiac Findings In Barth Syndrome

By Carolyn T. Spencer, M.D., University of Florida Pediatric Cardiology, Gainesville, Florida

Although a true long term study of patients with Barth syndrome has not been undertaken, we recently had the opportunity to evaluate the heart status of 19 boys who carry this diagnosis. The boys, ages 1-19 years (average 10 years) underwent echocardiograms, electrocardiograms (ECG) and a clinical questionnaire. In addition, genetic analysis was available on all patients.

All boys underwent an echocardiogram using a specific protocol designed for this study. This was aimed primarily at determining the size of the left pumping chamber (left ventricle, LV) and several elements of ventricular function (how well the heart contracts and relaxes). The size of the left ventricle was evaluated by determining the largest width and volume of the chamber and by estimating the mass (weight) of the chamber. Analysis revealed that as a group, the dimension and volume were upper limits of normal when compared to normal children for each age. Evaluation of the thickness of the left ventricle wall and mass revealed that the wall thickness was normal and the mass was normal for the size of the chamber for the group as a whole. Although some patients with Barth syndrome are known to have abnormalities in the thickness and appearance of the muscle in the lower portion of the LV (LV "non-compaction"), we did not see any significant "non-compaction" in this group. Multiple parameters of function were studied. The ejection fraction (EF) is a number that represents the percent of blood the left ventricle pumps out with each beat, with normal being about 55 – 75%. The average EF for this group of Barth syndrome patients was 49 % (range 22 – 62%). The shortening fraction (SF) is the percent change in the width (dimension) of the left ventricle with each cycle of contraction and relaxation, with normal being about 28-40%. The SF is usually a little more than ½ of the EF. In this group of patients, the average SF = 27 % (range 14 – 34%). Both of these elements are consistent with mildly reduced contractile function of the heart for the patients as a group, but with a wide range from severely decreased to normal. Other measures of ventricular function were evaluated and correlated with the assessment that the global heart function is mildly reduced in this group of patients. There was no significant relationship between the LV function or volume with age of the patients.

Analysis of the ECGs showed that some patients have evidence of increased LV size (as expected). The heart rhythms were normal and no abnormalities of conduction (electrical pathways in the heart) were detected.

Clinical questionnaires were available on 15 patients. Most patients were on heart medications (such as digoxin, lasix, and an ACE inhibitor such as captopril or lisinopril). All of the patients who completed the questionnaire describe below or far below average exercise tolerance compared to their peers.

Genetic analysis available on all patients was kindly provided by Dr. Iris Gonzalez. In 18 patients, there were many different mutations in the *G4.5* gene on the X chromosome, which has been identified as the "Barth" gene. One patient who was felt to have the clinical picture of Barth syndrome did not have a defect in the "Barth" gene. There was no identifiable correlation between the specific type or location of the mutation within the gene and the cardiac status.

In summary, several important points should be made from this analysis. Evaluation of the function data suggests that there is no progression of the cardiomyopathy with age, as there was no correlation of the EF or SF with age. The only way to confirm this is to follow a group of patients over time and see if the cardiac function changes with age. If there is no evidence of progression of cardiac disease, then this would suggest that other modifying factors influence

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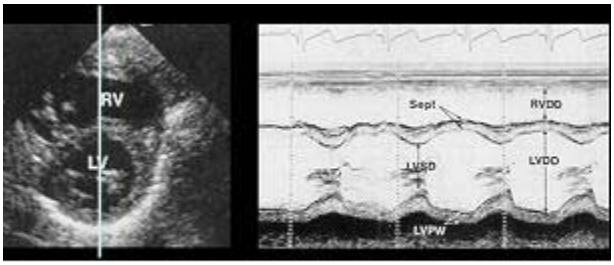
Cardiac Findings In Barth Syndrome



2-D and M-mode echocardiogram to evaluate cardiac function.
EF= (Dias. Vol. – Sys. Vol.) / Dias. Vol.
SF= (LVDD-LVSD)/LVDD

which patients with Barth syndrome have severe cardiomyopathy versus those who have a milder cardiomyopathy. Some of these factors could possibly be identified through a broad range of genetic testing by evaluating related genes. Although the contractile function was only mildly reduced in this group of patients, it should be remembered that most patients are on heart medications and under the care of a pediatric cardiologist. It is also possible that the 19 patients who were studied at this conference are not necessarily representative of the group of Barth syndrome patients as a whole. We can not be sure if only the healthier (or sicker) patients attended the meeting. Thus, it is important to include a larger group of patients in order to answer more questions about the heart function in Barth syndrome. We were not able to correlate cardiac function with any blood analysis, such as the white blood cell count. We also did not do any exercise testing, which may help determine how much of the exercise tolerance is due to abnormal skeletal muscles versus the decreased heart function.

We tremendously enjoyed meeting the wonderful families and boys who attended the Barth Syndrome Foundation conference. We look forward to continuing working with the Barth Syndrome Foundation and hope to be able to provide even more information in the future. Should a family who has a son with a confirmed diagnosis of Barth syndrome who did not participate in the Cardiology Research Clinic which was held at BSF's 2002 conference wish to participate in this study, please contact Dr. Spencer at the following address: Carolyn Spencer, MD, University of Florida, Pediatric Cardiology, Box 100296, Gainesville. FL 32610 (Phone: 352-392-6431).

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American Heart Association Awards Grant in Support of Barth Syndrome Research

We hope to find answers to these questions within three years, the period for which this grant is funded. But even under the best of circumstances it would be impossible to do so without the help of dedicated scientists. That is why we are pleased to announce that Dr. Yang Xu has agreed to join our team to do research on the mechanism of Barth syndrome. Dr. Xu has extensive experience in biochemistry, cell biology, and molecular biology. She received an M.D. from Jinzhou Medical College in Jinzhou, China, and a masters degree in biochemistry from The Fourth Military Medical University in Xian, China. She then advanced her professional skills in several top-ranking US institutions, including Columbia University, Mount Sinai School of Medicine, and Albert Einstein College of Medicine in New York.

Step by step we hope to expand our knowledge of Barth syndrome. Dr. Xu's work, as well as the research of other groups funded by the Barth Syndrome Foundation, will eventually result in a better understanding of this disease. It will guide us in our efforts to design more efficient treatments for children with Barth syndrome and perhaps also for children with other cardiac and muscle diseases.

2004 Barth Syndrome International Family and Scientific Conference

By Anna Dunn, Vice President and Family Liaison

Coronado Springs Resort
Lake Buena Vista, Florida
July 8-12, 2004

On behalf of the Barth Syndrome Foundation, Inc., (BSF) I would like to personally invite you to attend the ***2004 Barth Syndrome International Family and Scientific Conference (hereinafter referred to as "conference"),*** scheduled for July 8-12, 2004 at the Disney Coronado Springs Resort in Lake Buena Vista, Florida. This conference will provide the most up-to-date information on the forefront of research and resource material on Barth syndrome. All scientists, researchers, educators and Barth family members (including siblings and grandparents), and other interested individuals are invited to attend.

The *Scientific and Medical Sessions* will include sessions on both the clinical aspects of Barth syndrome as well the underlying basic science of the disorder. Scientists and researchers interested in many facets of Barth syndrome will collectively share their knowledge as it pertains to this disorder.

The *Family Sessions* will consist of various panel discussions of Barth topics including neutropenia, cardiology, neurology, hematology, and education. Along with these sessions, individual clinics for the Barth boys will be offered.

Presentations: Expert and involved medical-scientific participants will present on specific Barth syndrome topics such as cardiology, hematology, and neurology with basic genetics, biochemistry, and mitochondrial bioenergetics and gene therapy, amongst others.

Panel Discussions: Various medical-scientific participants will focus on several specific areas of Barth interest. The specialists will open with a brief introduction of their specialty, followed by an interactive "Question and Answer" forum between the panel of specialists and the Barth families.

Round Table Discussions: Interaction amongst Barth families, under the supervision of a moderator, which will focus on Barth-related issues and concerns as it pertains to this disorder.

BSF has reserved a limited number of rooms on a first call, first serve basis at a special room rate of $115.00 (plus tax) per night, so be sure to **reserve your room at #407-939-1020 any time after July 8, 2003**. The Coronado Springs Resort is situated around a 15-acre shimmering lake, "Lago Dorado". The resort offers vast walking nature trails, 5 themed pools with water slides, hot tubs, themed playgrounds, and most importantly it is wheelchair accessible. Free bus transportation for all Disney theme parks is readily available at the front entrance.

If you have any questions in regards to our upcoming conference, do not hesitate to contact me, Anna Dunn, Vice President and Family Liaison, at adunn@barthsyndrome.org. Please go directly to BSF's website at **www.barthsyndrome.org** and **fill out** the ***2004 Barth Syndrome International Family & Scientific Conference Registration Form*** as soon as possible**,** and be sure to frequently visit our website for current updates.

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Important steps to take prior to the 2004 Barth Syndrome International Family and Scientific Conference:

1. Fill out the Registration form located at the BSF website, **www.barthsyndrome.org** ASAP.

2. Reserve your vacation time for the upcoming 2004 conference.

3. Call the Coronado Springs Resort any time after July 8, 2003 at **407-939-1020** (from all locations) and reserve your room at $115.00 (plus tax) special rate. Rooms will be given on a first call, first serve basis…so mark your calendars for July 8, 20003 to reserve your room!! Be sure to mention that you are reserving rooms for the ***"2004 Barth Syndrome International Family and Scientific Conference"***.

4. Barth families, ask your son's physicians to reserve their schedule in advance if they are interested in attending the upcoming 2004 Barth Syndrome Scientific portion of this conference, which will be held in conjunction with the 2004 Barth Syndrome family meetings. Dr. Richard I. Kelley will be hosting the scientific portion of this conference, and all interested physicians and scientists are invited to attend these meetings. Most of the scientific meetings will run separately along side with the family meetings. Some of the sessions will be held jointly with the Barth families so that all will have the opportunity to meet together.

This upcoming conference is a golden opportunity to obtain the most-up-to-date information on Barth syndrome from various distinguished Barth syndrome scientists/researchers/educators, as we unite as "one family." Barth children, siblings, parents and other family members will all have the opportunity to share their piece of the Barth puzzle. Together we can all visualize the hope for a better tomorrow for our Barth children and future generations, so that one day, we can confidently say, that ***"...not one more child will suffer or perish from this condition."***

Why Do We Need A Barth Syndrome Disease Registry?

By Gerald F. Cox, M.D., Ph.D.
Medical Director, Genzyme Corporation, Cambridge, MA
Division of Genetics, Children's Hospital, Boston MA

Before attempting to answer this question, let me first explain what a disease registry is and what it is supposed to do.

What is a Disease Registry?

A disease registry is an organized way of collecting, storing, and analyzing clinical information on a patient population over time to gain a better understanding of the disease. Disease registries are useful for identifying important disease-related trends, detecting rare side-effects associated with new drugs, and for assembling comprehensive information on rare diseases. In addition to collecting disease information, some registries have affiliated repositories that store blood samples, tissue samples, or cell lines that can be made available to researchers for future studies.

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(Continued from page 12) **...Barth Syndrome Disease Registry**

How Does a Disease Registry Work?

Patient participation in disease registries is often voluntary, although some disease registries are maintained for public health reasons (e.g. SARS or AIDS) or required by regulatory agencies to monitor the long-term safety and efficacy of new drugs. Once a patient and/or legal guardian agrees to enroll in a disease registry and signs an informed consent form, a case report form (CRF) is filled out by a health care provider (physician, nurse, or study coordinator). A CRF allows patient information or data to be collected in a systematic manner so that each piece of data can be summarized and compared across patients. Data fields on the CRF are chosen with input from disease specialists, who may constitute a board of advisors. Paper CRF are still used most commonly, but electronic data capture forms, in which the data is typed into specified fields on the computer and then emailed instantly to the disease registry, are becoming more popular. Patient confidentiality is protected by assigning a patient code or number on each CRF in place of the patient's name. Disease registries require an infrastructure consisting of one or more dedicated individuals involved in the design, implementation, and maintenance of the registry as well as financial resources.

How is a Disease Registry Used?

For rare diseases, a disease registry has the potential to literally define the disease, including its clinical features and spectrum of severity, by summarizing data on large numbers of patients from different geographic regions. A disease registry is able to better characterize the variability, progression, and natural history of disease than anecdotal reports of individual patients. These data can be used to show trends over time in diagnosis, illnesses, disability, treatment, and outcomes. The summary information can be used to help develop and substantiate guidelines for monitoring and treating patients. In diseases where a clinical study involving the use of a placebo control group would be considered unethical, a disease registry can provide important information on untreated patients. Registries can evolve over time as new clinical developments arise by simply adding new data fields. Working with registry staff and the board of advisors, outside clinical researchers can query the registry to test their hypotheses.

Why Should a Patient or Physician Participate?

Everyone benefits from participating in a disease registry. Patients who are enrolled in the disease registry will have the personal satisfaction of knowing that their data will help to advance patient care. Physicians and other interested parties will become better educated about a disease and its treatment through medical journal articles and other publications based on disease registry data. An individual physician can compare how his or her patient is doing relative to the larger patient population. A disease registry also provides a valuable resource for academic clinicians who think about the patients to make important scholarly contributions to the field by describing new clinical aspects of a disease, testing hypotheses, or describing outcomes with and without certain interventions or treatments.

Back to my question "Why Do We Need a Barth Syndrome Disease Registry?"

I'll answer this question with another: "Why not?" Barth syndrome is a rare genetic disease, an "ultra-orphan" by any standards (by definition, "orphan diseases" affect fewer than 200,000 individuals in the US), that would benefit greatly from having its own disease registry. With fewer than 100 known patients, a central repository for clinical data would provide a valuable resource for clinical researchers. Clinical data provided by small subsets of patients are simply incapable of adequately describing the characteristics of the entire patient population. Only with a critical number of patients is it possible to know what is common and what is not, what is expected and what is not, and what works and what does not.

Our understanding of the underlying basis of Barth syndrome has proceeded at an amazing pace over the past few years. Within a short timeframe, we have learned of the genetic cause of Barth syndrome, and we are getting close to understanding the biochemical basis of the disease. Research grants provided by the Barth Syndrome Foundation to several scientists this year will accelerate the pace of future discovery. An equally important role of the Foundation is to provide emotional support and information to families afflicted with Barth syndrome from across the world. A strong network of friends has developed that is dedicated to improving the lives of others facing similar circumstances.

(Continued on page 14)

(Continued from page 13) **...Barth Syndrome Disease Registry**

Although mechanisms are in place to guide future laboratory research and provide support to families, what about the patients themselves and our understanding of the medical aspects of Barth syndrome? What can we clinicians do now, not 5 to 10 years down the road, to improve the health and lives of patients with Barth syndrome? I strongly believe that a Barth Syndrome disease registry is what's missing from the big picture, and that such a registry can be a major driving force that impacts future patient care, potentially to the same degree that the awarding of scientific research grants will enable future discoveries. To be successful, though, a registry requires commitment and cooperation from all involved parties. There needs to be financial support for the registry infrastructure, either from the Foundation itself or from a granting agency. Patients and their families must be willing to enroll into the registry and contribute demographic and health-related information on an ongoing basis. Health care providers must take time from their busy schedules to enter patient information. A board of advisors needs to provide medical guidance and oversight to the registry. Finally, there needs to be registry staff members to design the CRF, set up a computerized database, analyze the data periodically, and with the board of advisors, publish the findings.

The time is now and the choice is ours. I've made my thoughts known, but I'd like to hear what the other members of the Foundation think. I can be contacted at **gerald.cox@genzyme.com** or at Genzyme Corporation, One Kendall Square, Cambridge, MA 02139.

2002 BSF Financial Report

By Steve McCurdy, Vice President, Finance and Development and Michael Wilkins, Treasurer

Following this report are a summary statement of Revenue and Expenditures and a Balance Sheet for the Barth Syndrome Foundation, Inc. for the fiscal year ended December 31, 2002. As required by the IRS and many of the 10 states in which we are registered, our financial statements have been audited by Buckley, Sitzman and Nielsen CPAs in Lincoln NE. You will also be able to find our financial statements on our website **www.barthsyndrome.org**.

BSF is a strong and financially healthy organization due to the efforts of a growing group of dedicated volunteers and a very active Board. We exceeded our objectives in every area in 2002. We raised over $328,000 in 2002 vs. $145,000 in 2001, and finished the year with total assets of over $418,000. We also had awarded but not yet paid some $77,000 in research grants and together with other accounts payable left us with $341,000 in net assets at year-end 2002. A charitable organization like BSF must distinguish between assets it holds that are restricted by the terms set by a donor, and those that are unrestricted. BSF held a little over $29,000 in funds that were restricted to research use only, and over $312,000 in funds with no restrictions whatsoever.

Our expenses were predominately in support of program services. We spent almost $112,000 in this area in 2002, principally on research grants. We spent a little over $14,000 on administrative and general expenses and almost $6,600 on fund raising. Many private and governmental groups rate charities on their fund raising effectiveness by looking at the ratio of fund raising expenses to funds raised. Our ratio of 2% in this area is excellent. Likewise, charities are rated on the ratio of program expenses to total expenses. In this area also, our ratio of 84% is also excellent.

Finally, we can report that we passed our audit with flying colors. Contributors can be confident that BSF utilizes professional bookkeeping, control, planning and investment tools and that our programs are carefully planned and managed to accomplish our goals most efficiently. Although we remain an all-volunteer organization, we strive to learn from the best practices of other organizations and take our responsibilities seriously.

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Barth Syndrome Foundation, Inc.
Statement of Financial Position
as of December 31, 2002

	2002	2001
Assets		
Current Assets	$ 385,522	$ 39,563
Other Assets	$ 29,597	$ 102,341
Total Assets	$ 415,119	$ 141,904
Total Liabilities	$ 76,899	$ - -
Net Assets		
Unrestricted	$ 312,182	$ 40,618
Temporarily Restricted	$ 29,038	$ 101,286
Total Net Assets	$ 341,220	$ 141,904
Total Liabilities and Net Assets	$ 418,119	$ 141,904

Barth Syndrome Foundation, Inc.
Statement of Activities
For the Year Ended December 31, 2002

	2002	2001
Support and Revenue		
Contributions	$ 328,344	$ 145,327
Other	$ 3,682	$ 1,806
Total Support and Revenue	$ 332,026	$ 147,133
Expenses		
Program Services	$ 111,808	$ 3,281
Management & General	$ 14,331	$ 2,162
Fund Raising	$ 6,571	$ 483
total Expenses	$ 132,710	$ 5,926
Change in Net Assets	$ 199,316	$ 141,207
Net Assets, Beginning of Period	$ 141,904	$ 697
Net Assets, End of Period	$ 341,220	$ 141,904

$500,000 Contribution for Research Highlights Growing Support for BSF

By Steve McCurdy, Vice President, Finance and Development

It is a tribute to the hard work and dedicated efforts of hundreds of volunteers – families, friends of families and those who have been inspired by our passion to solve this puzzle called Barth syndrome. It is in recognition of the high degree of professionalism and stewardship with which BSF has handled previous contributions and especially the success we have had in assembling a world-class Scientific and Medical Advisory Board and creating a research grant process for grants to be awarded by the Barth Syndrome Foundation. And finally, it is in honor of Kevin Baffa and the Baffa family – a family of faith and friends and strong supporters of the hopes and ideas behind BSF since our first informal conference in Baltimore in 2000.

"It" is the strongest vote of confidence that BSF has received yet, and a confirmation that we are on the right path toward finding a deeper understanding of the underlying causes of Barth syndrome and ultimately a cure for this genetic, metabolic puzzle. "It" is a $500,000 contribution to BSF to be used to fund research grants over the coming years and a major milestone in the young life of the Barth Syndrome Foundation.

At the request of the contributing foundation, its identify will remain confidential. But the story behind this incredible gift goes back a few years. In 2001, an anonymous foundation whose leaders had taken an interest in Barth syndrome and BSF generously gave us a $100,000 gift to be dedicated to funding research. The problem was that BSF had just been formed and had no way to reasonably attract, let alone sort through and select the best research projects to fund. Rather than rush ahead into a world for which it was not prepared, the Board decided it needed help and assigned Kate McCurdy to build a Scientific and Medical Advisory Board and a grant application, review and award process. The scientific experts and the grant review process were meant to insure that BSF research "investments" would be of the highest quality and most likely to lead to improvements in our understanding of the causes of Barth syndrome and to improvements in treatment and care for Barth boys.

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It took the start of 2002 before the SMAB could be assembled, with the grant process designed and approved by the SMAB and the BSF Board by early 2002. This edition of the BSF Newsletter reviews the results of that process with the first awards of five high quality scientific research projects. The funds for these projects come largely from that initial leap of faith by our anonymous friend and benefactor.

But our benefactor was not through yet. Seeing the methodical and careful process that BSF went through to make sure that we would use their research funding wisely, they encouraged us still further with an additional $100,000 grant in 2002… this time the contribution was un-restricted and could be used to kick off our awareness programs and family support efforts such as the Listserv.

But it was the continued efforts of the Barth syndrome families and their friends to raise money and their success in creating a broad base of concerned contributors that convinced our anonymous benefactor that BSF would not allow itself to become dependent on a single contributor. In 2002, Gary Rodbell raced in the Ironman Triathlon for BSF and raised over $79,000; Lynda Sedefian led her 2nd Walkathon which raised over $14,500, the Kuglemann's Barth Syndrome Golf outing raised over $20,000, the Mann's car rally raised over $3,000 and the Wilkins and the McCurdys again sent out their annual solicitation letters to their friends and families. Each of these efforts proved very effective in raising money *and* awareness for Barth syndrome in 2002.

Bill Fagan

In 2003, the amazing friends of the Baffas, led by Bill Fagan have already held a wonderfully fun and heart warming dinner-dance-auction to raise money for BSF, and the Dussich Dance Studio where R. J. Kugelmann takes an acrobatics class, contributed a part of the proceeds to BSF from their March "Sharing through Dance" show . And BSF received its first long term commitment from a contributor who pledged $5,000 per year to BSF "until a cure is found". This year has gotten off to a strong start and we look forward to more fund raising efforts to come.

All of these efforts give BSF credibility and demonstrate that the combined passion and sheer positive force-of-will of the families that form the core of BSF will insure our ultimate success.

With the support of our growing base of families and volunteers, our cadre of caring contributors, our dedicated SMAB and our anonymous benefactor, how can we NOT succeed?

On behalf of the Board, the Baffas, the boys and all of us who care, dear benefactor…and all of our contributors… **We Thank You**!

From left to right: Steve McCurdy; Richard I. Kelley, M.D., Ph.D.; Kate McCurdy; Shelley Bowen; Rosemary Baffa; Ted Baffa

BSF Hosts Steinhatchee Workshop

By Michaela Damin, President
The Barth Syndrome Trust - United Kingdom

First Row (L-R): Sue Wilkins, Cathy Ritter, Shelley Bowen, Alanna Layton, Chris Hope, Rosemary Baff

2nd Row (L-R): Karen Gordon, Michaela Damin, Lynda Sedefian, Shelia Mann, Holly Edsel, Lynn Elwood

3rd Row (L-R): Steve McCurdy, Kate McCurdy, Anna Dunn, Mark Dunn

Top Row (L-R): Steve Kugelmann, David Mann, Mike Wilkins, Michael Bowen

When I was invited to attend the 2003 Workshop in Steinhatchee, Florida, I was thrilled for many reasons. Here was an opportunity to see my friends again, friends I may have only met once before at the 2002 Conference but who nonetheless have the dubious honour of now being considered part of my family. On a more serious note, I had been feeling for some time that I wanted to play a more active role in helping each and every one of our boys and their families. The only problem was that I didn't know where to start! Upon talking to other families, it became evident that there were a number of us who felt the same way.

Just as we were realising this, along came our invitations! Our erstwhile leaders saw the workshop as an ideal opportunity to plan for the future of the group. A great deal of planning had originally gone into devising BSF's vision, mission and program goals in September 2000. Two and a half years later, it was apparent that the foundation laid was sound. However, as we continue to grow in strength and numbers, we need to regularly re-evaluate our goals. In doing so, we can ensure that we, as an organisation, are always on track and serving each and every one of our members in the best possible manner.

I'm sure I'm not alone when I chat to friends and family and say, "Isn't it incredible how much we have accomplished in such a short space of time?" But I never really included myself amongst those actually doing the accomplishing! Whilst we are always so grateful to our "superheroes" for all they do, we are also always aware that every moment they spend trying to save our boys is a moment they spend apart from their own boys and family.

And so, we began to develop comprehensive plans to share some of these tasks. Shelley Bowen, Steve and Kate McCurdy, Sue and Mike Wilkins and Anna Dunn each spoke about what they have currently been doing and then highlighted

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BSF Hosts Steinhatchee Workshop

WATCH your newstand for the June 2003 issue of Reader's Digest which will be featuring an article about Barth syndrome and Shelley Bowen, our President, entitled *"Saving Michael Bowen"*. Please be sure to pick up a copy of this issue.

specific areas in which they would benefit from assistance. The "Wish Lists" which each person created provides us with a breakdown of individual tasks, many of which are vital but which they understandably have been forced to delay. Many of us at the workshop realised that suddenly here was a task that interested us personally that we could tackle at our own pace, with the aid of all the board members and other "seasoned veterans".

We are all the same - we each struggle with coping with our boys and their health, our families, our jobs, our households. If you would like to find out more about how you can help, PLEASE contact me (mdamin@barthsyndrome.org) and I will gladly chat with you and send you copies of the Wish Lists.

Another recent development has necessitated a detailed planning session and that is the article about Barth syndrome which is due to appear in the June issue of the Reader's Digest. With 90 million readers in 19 different countries, we needed to be prepared for the possibility that we could receive an overwhelming response. We now have specific people assigned to dealing with enquiries from diagnosed and undiagnosed families, from physicians and scientists, from the media, from people wanting to help and from people needing our help. Our aim is to respond quickly and efficiently to each enquiry. This is a wonderful opportunity for us and we are now prepared for it!

Another topic on the agenda was the Registry. I have long dreamed of the day when I could have a comprehensive set of data on my own son. We have already learned so much from just sharing information amongst ourselves. As parents, we sometimes notice small details and sometimes overlook others but it is always exciting when others say "That's strange, my boy does exactly the same thing, or eats the same foods or has exhibited similar symptoms!" Conversely, we can learn so much from the differences between us, as each of our boys are unique. From the data gathered, it is my hope that we will take the first step towards answering the question "Why?" - "Why is my son similar?", "Why is he different?" Patterns may emerge, myths may be dispelled; at the very least, keen interest will be enerated. The data compiled should prove to be of tremendous value to us, as carers but also to the physicians and scientists researching this disorder who are helping us care for our boys.

Another exciting event that requires a huge amount of planning is the 2004 International Family and Scientific Conference to be held at the Coronado Springs Resort, Lake Buena Vista, FL. We will be paying special attention to the needs of "first-time" families; we plan to include research updates, panel discussions and clinics, to name a few. We also plan to have fun! Our son, who does of course rule the household (as well as the budget!) has already decreed that we will be going. He is also inviting all his friends! I can't wait to see you all there; I look forward to seeing old friends and to making new ones.

There was so much to cover and plenty to do, and of course, every spare moment was crammed with parents talking to each other. "What is this connection we have?" It transcends boundaries and borders and it gives us strength and unity and purpose. We laughed (my jaw still aches!) and sometimes we cried a little and we all worked together. And it worked!!

Articles Relevant to Barth Syndrome Published in Professional Journals Since Last Newsletter

Strauss A, Lock JE. **Pediatric cardiomyopathy – a long way to go.** Editorial. N Engl J Med 2003 April 24; 348(17):1703-1705.

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Folger U, Cario H, Panis A, Keck T. **Postinflammatorial stenoses of the pharynx in a child with Barth syndrome.** Int J Pediatr Otorhinolaryngol 2003 Feb; 67(2)117-120.

Vesel S, Stopar-Obreza M, Trebusak-Podkrajsek K, Jazbec J, Podnar T, Battelino T. **A novel mutation in the G4.5 (TAZ) gene in a kindred with Barth syndrome.** Eur J Hum Genet 2003 Jan; 11(1):97-101.

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Chen R, Tsuji T, Ichida F, Bowles KR, Yu X, Watanabe S, Hirono K, Tsubata S, Hamamichi Y, Ohta J, Imai Y, Bowles NE, Miyawaki T, Towbin JA. **Mutation analysis of the G4.5 gene in patients with isolated left ventricular noncompaction.** Mol Genet Metab 2002 Dec; 77(4):319-325.

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Barth Syndrome in Many Languages!

By Lynn Elwood, Vice President
The Barth Syndrome Foundation, Canada

It has become clear that Barth syndrome is a disorder that affects boys around the world. We now know about families in many countries around the world, and with all types of backgrounds. As the Reader's Digest article helps us to reach out to all areas of the world we have a new challenge to face – many families may want information in languages other than English.

From left to right: Dorian Hileaga, Victoria Nirenshtein, Meenakshi Joshi-Butail, Kevin Zhong, and Aleksandar Susnjar

Languages: Romanian, Hindu, Chineese (2 styles); Russian; Hrvatski; Cirilica; and Bosnia

Recognizing this need for help in other languages, Shelley requested assistance to translate a key web-site page that tells about Barth Syndrome symptoms and how to reach the Barth Syndrome Foundation. Through the Listserv, many families responded and over a few short weeks several translated pages were supplied from areas all around the world. We now have the page translated into 15 languages and have ambassadors to help with inquiries in most of those languages. What an incredible effort this has been.

I have the privilege of working with a very multicultural group of people in Toronto, Canada. When I mentioned the need to translate the page to different languages, the group I work with sprang into action and within hours started sending me translated pages to pass along to the BSF. It brought tears to my eyes to see that first translated page and the out-pouring of support these people provided. They were, and continue to be very quick to help the BSF and the boys in any way they can. Top left is a picture of the amazing group from Toronto who provided some of those initial translations. Thanks to them and to all the other families and BSF friends we now have the chance to reach families all over the world and help them to find out about this disorder in their language of choice. Thanks to all of you!

Annick Manton ~ French

Iris Gonzalez ~ Spanish

Nori Damin ~ Italian

Joke vanLoo ~ Dutch

Helena Freündt ~ Swedish

The Power of Kindness

(Contributions Received Since May 2002)

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Growing up with Barth Syndrome

By Robert Hope, Canada

Despite the fact that I have Barth syndrome, I do try to fit in with the people at my school and lead the life of a "normal" seventeen year old. Although I don't play sports, I was part of the school's archery team. I have also been taking part in the school drama courses and the junior drama club. Because of my light build and weight, I was even offered a position on the school wrestling team. Of course my mother was a little uncomfortable about that. Outside of school I enjoy swimming and fishing in the summer, and I even took horseback riding lessons.

Normally whenever I mention the disorder, people don't know what I'm talking about. I am happy that this is one of the times where I don't have to explain what it is and how it affects me. Whenever I try to explain to people when they ask about it, they just don't understand. Whenever I say Barth syndrome to them, they think I say Barf Syndrome! (Maybe we boys should start calling it something else—any suggestions?). Even though I have friends at school that realize that I can't do things that they can do and they respect me by not saying anything about it, it is even better knowing other people that also have Barth syndrome. Sometimes if I feel rejected I think of the friends that I made at the conferences and they give me hope and it makes me feel better.

I try to cope with the disorder as best I can but at times I do feel like it's an endless struggle for survival. I know I am actually one of the lucky boys that have this disorder. Some of the boys can only get through one or two periods of class a day while I can almost manage a full curriculum, and some are sick on a regular basis, which I am not.

I enjoyed both of the conferences because they allowed some of the families to get together, and us boys to share experiences with each other. Finally I wasn't the only one in a group that had this, I was with boys my own age that were the same. It felt great to be "one of the guys", listening to music, playing chess, pool, and not having to be worried about being singled out.

After meeting some of the doctors, including Dr. Barth, at the conferences I am truly amazed at how much they actually understand about the disorder and how easily they explained it to us. I am already looking forward to seeing everyone again at the next conference…it will definitely be a blast!

Saving boys' lives through education, advances in treatment and pursuit of a cure

Barth Syndrome Foundation

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