



The BSF Newsletter

Introduction from BSF's President, Shelley Bowen

Dear Friends,

I cannot begin to put into words how amazing it is to address you in our first newsletter. This is a great accomplishment and my heartfelt thanks go to Lynda Sedefian for her tireless effort in making this newsletter a reality.

Just over eleven years ago I heard the term Barth syndrome for the first time. Like so many of you we were, as parents, facing this battle alone. We had no idea what we were facing. This newsletter you hold in your hands marks a historical accomplishment. This newsletter gives new found hope to those we know and those we have yet to find. It is the mission of The Barth Syndrome Foundation to find undiagnosed individuals and assist those of whom we are aware. Far too many children's lives have been claimed and far too much suffering has taken place as a result of the disorder our children bear. There is now more knowledge about Barth syndrome and never again should any family feel the sense of isolation, which so many of us have experienced in the past.

I often think back to Baltimore just over a year ago. I think of the families sitting on the floor discussing concerns and the desire to move this group beyond a group of parents and into a realm of an advocacy group of which
(cont'd on page 2)



Valerie (Shelley) Bowen, Pres., BSF

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Walk-a-thon a Huge Success

Lynda Sedefian raised over \$16,000 by putting on a walk-a-thon in her hometown of Voorheesville, New York.

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Photograph submitted by Perry Shillinglaw, Voorheesville, New York

Barth Syndrome Now Recognized by NORD

Vision...A world in which not one more child will suffer or perish from Barth Syndrome.

Mission...To guide the search for a cure, to educate and support physicians and to create a caring community for affected families.

For the first time ever, NORD has recognized Barth syndrome within their database of rare diseases, a huge milestone achieved by the Barth Syndrome Foundation, Inc. (BSF). As a result, BSF approached Gerald F. Cox, M.D., Ph.D., F.A.C.M.G, Medical Director, Department of Clinical Research, Genzyme Corporation, Cambridge, MA 02139; Assistant in Medicine, Division of Genetics, Children's Hospital, Boston, MA 02115, seeking his expertise to prepare the following article which will be incorporated into The NORD Guide to Rare Diseases. This article is printed with the permission of the National Organization for Rare Disorders; Publisher: Lippincott, Williams & Wilkins; In Press, 2002; Copyright belongs to NORD, which reads in its entirety:



Gerald Cox, MD, Ph.D., F.A.C.M.G.

Barth Syndrome

DEFINITION: Barth syndrome is a rare X-linked genetic disorder characterized by the triad of cardiomyopathy, neutropenia, and 3-methylglutaconic aciduria in boys. The disease gene, G4.5, encodes a group of related proteins called tafazzins that are involved in the metabolism of phosphatidylglycerol and cardiolipin. In addition to Barth syndrome, mutations in the tafazzin gene may cause isolated dilated cardiomyopathy, left ventricular noncompaction, endocardial fibroelastosis, and cyclic neutropenia.

SYNONYMS: X-linked cardioskeletal myopathy and neutropenia, endocardial fibroelastosis (EFE) type 2; 3-methylglutaconic aciduria type II

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**FIRST-EVER BARTH SYNDROME
FAMILY/PHYSICIAN CONFERENCE HELD
IN BALTIMORE, MARYLAND IN JUNE OF
2000**

In June of 2000, families from four different continents gathered at the Mt. Vernon Hotel in Baltimore, Maryland to gain more insight into the nature of Barth syndrome, as well as to connect with other families who live day-to-day with this rare genetic disorder. In attendance at this conference were a number of very well-informed physicians, geneticists and researchers who shared very valuable information in terms of the most recent data on Barth syndrome. To name a few, there were Peter G. Barth, M.D., University of Amsterdam, Amsterdam, The Netherlands who focused on "X-linked cardiomyopathy and neutropenia - an historical and clinical overview of Barth Syndrome"; Dr. Richard Kelley, M.D., Ph.D., Kennedy Krieger Institute, who focused on "Barth syndrome, the American Experience", "Organic acid metabolism in Barth syndrome", and "Cardiomyopathy-Parkinsonism syndrome: an autosomal recessive Barth-like disorder"; Gerard Berry, M.D., Children's Hospital of Philadelphia, who focused on "Treatment of Barth syndrome with growth hormone"; Gerald Cox, M.D., Ph.D, The Children's Hospital, Boston, MA who focused on "Neutropenia in Barth syndrome: treatment with G-CSF"; Iris Gonzalez, duPont Children's Hospital, Wilmington, DE, who focused on "The spectrum of G4.5 mutations in Barth syndrome"; Troy Phipps, Doctoral Candidate, California State University, Northridge, who focused on "Subcellular Localization of the Barth Proteins"; and Peter Vreken, Ph.D., University of Amsterdam, who focused on "Phospholipid abnormalities in Barth syndrome".

The Barth Syndrome Foundation is in the midst of planning for our 2002 family/scientific conference which will be held in Baltimore, Maryland in October of 2002.

Please watch your mailboxes for information pertaining to BSF's 2002 conference. Anna Dunn has agreed to take on the responsibility of Chairperson for this conference and any questions should be addressed to Anna at: adunn@barthsyndrome.org

We hope to see you there!

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Introduction from BSF's President...

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to take notice. I am reminded of the emotional sentiment shared by all, isolation. Isolation encompasses a number of fronts. We were isolated from clinicians knowledgeable about this disorder, we had access to articles but no one to translate the scientific terms, we had little if any contact with parents in the same situation to field questions on seemingly insignificant details to others but crucial to our children. The first conference bridged many continents through a common bond. It was a beautiful experience. From different corners of the world and cultures apart we all came together for one common purpose. We wanted to learn more to ease the suffering of our children and future generations. We were making an investment into our future. We wanted to fight this thing together! The first conference offered a forum for emotional healing, a time to share tears with a friend we never met face-to-face, and an opportunity to embrace human touch never before known. Three mothers who had never met each other in person worked together to put this meeting together. We, like you, saw the need for this experience to unite together to combat this disorder as a group. I look forward to seeing those faces again and meeting the new families who have come to our group since that time as well as those who were not able to attend the first conference.

This past year I have had the privilege to work on your behalf with some of the finest people I have ever known. Each of our board members has selflessly devoted thousands of hours to this common cause. I am constantly amazed at how they all come from diverse backgrounds and bring to the table unique perspectives, but at the base, have the best interests of all of our children at heart. I know each of you join me in thanking them for their effort to make BSF a reality and no longer a dream of what could be.

If history has taught me anything it is that faith the size of a mustard seed can move mountains. Small-disenfranchised groups throughout history have changed the course of events through impassioned belief. Our champions throughout history were built cell by cell. As the cells combined they grew to be human beings, experiences they encountered along life's path revealed a mission. Our coming together personifies life, faith and hope. Each of us brings to this circle of wisdom a piece of this puzzle, a cell if you will. We have grown and we are moving steadfastly forward. BSF is our champion because it represents all of us. Our hero's are our children. We as a group are working side by side for a common goal...Not one more person should suffer or perish from this disorder.

Each of us has seen history re-write itself where this disorder is concerned. No longer do we call this a lethal disorder but a life-threatening disorder. We know more than ever before. Through connections with devoted researchers and clinicians we are learning more each day about Barth syndrome. Through advancements in science we have a greater hope than ever to see this life-threatening disorder become a treatable disorder. Our ultimate desire is to see this as a curable disorder.

So to each of you I say...Keep moving those mountains.

Shelley Bowen

President, The Barth Syndrome Foundation, Inc.

Barth Syndrome Now Recognized by NORD...

(cont'd from page 1)

DIFFERENTIAL DIAGNOSIS: Mitochondrial myopathy, including infantile MELAS syndrome (cardiomyopathy, myopathy, 3-methylglutaconic aciduria, respiratory chain abnormalities); myocarditis (cardiomyopathy); Kostmann and Shwachman-Diamond syndromes (neutropenia)

SIGNS & SYMPTOMS: Barth syndrome generally presents as heart failure in infant boys and rarely in older children. Initially, the cardiomyopathy has a distinctive dilated and hypertrophic pattern that often includes endocardial fibroelastosis. Over time the cardiomyopathy usually improves or resolves. At least three patients after a period of well-being experienced sudden cardiac arrest during childhood, presumably from arrhythmia. At least six patients have undergone heart transplantation, four successfully. Neutropenia may be cyclic or chronic and may predispose some boys to mouth ulcers and bacterial infections. Mouth ulcers may be associated with spiking fevers. Other variable symptoms include mild to moderate growth retardation, hypotonia, motor delay, and non-progressive weakness. Boys often have a myopathic facies and difficulty chewing. Cognitive function is intact. Historically, boys died of heart failure or infection by 3 years of age, but with improved diagnosis, medical therapy, and monitoring, survival has been extended into adulthood.

ETIOLOGY/EPIDEMIOLOGY: The Barth syndrome gene is located on chromosome Xq28 and is predicted to encode an acyltransferase. More than 30 mutations have been described in patients with no clear phenotypic correlation. The recurrence risk to future sons is 50% if inherited from an asymptomatic carrier mother, which accounts for 80% of cases with the rest being sporadic. The incidence may be as high as 1/100,000 with underdiagnosis likely. More than 50 families from North America, Europe, and Australia are known to exist through the medical literature and Barth Syndrome Family Support Group.

DIAGNOSIS: The diagnosis should be suspected in any infant boy with dilated cardiomyopathy. The diagnosis is confirmed by a high level of 3-methylglutaconic acid (2-20x normal) in plasma or urine and/or a mutation in the tafazzin gene. Bone marrow biopsy shows an arrest at the myelocyte stage of neutrophil development. Additional laboratory findings include monocytosis, intermittent lactic acidemia, hypocholesterolemia, abnormal appearing mitochondria in cardiac muscle, partial deficiencies in mitochondrial respiratory chain enzymes, and elevated levels of urinary 3-methylglutaric and 2-ethylhydracrylic acids. However, mitochondrial abnormalities vary in severity and sometimes are absent. Cultured patient fibroblasts exhibit low cardiolipin levels and reduced incorporation of linoleic acid into cardiolipin and phosphatidylglycerol (Vreken pathway).

TREATMENT: Standard Therapy: Medical care is supportive, and physical therapy is beneficial. Cardiomyopathy, which typically is most severe during infancy, is treated with standard medications, including cardiac glycosides, afterload reducers, and diuretics. Periodic monitoring for arrhythmias is indicated. Young boys should be observed closely for signs of infection as inflammation is reduced when neutropenia is present. In the setting of fever and neutropenia, young children should be evaluated for sepsis and treated with IM/IV broad-spectrum antibiotics until cultures are negative. Daily G-CSF SQ restores neutrophils to a normal level after a few days but the effect is transient without regular dosing. G-CSF may be used acutely or chronically to treat recurrent infections as well as mouth ulcers and gingivitis that are refractory to good dental hygiene and antiseptic mouthwash. Some infants have received antibiotic prophylaxis. Despite lifelong neutropenia, the rate and severity of infections decrease with age. Vaccinations and normal school participation are encouraged. No specific diet is recommended at this time. Although most children experience steady improvement after diagnosis during infancy, transient regression may occur during puberty.

Investigational: Some patients with failure to thrive may respond to pancreatic enzyme supplements. Vitamin supplements (Vitamin C, Vitamin E, Co-Enzyme Q10, and carnitine) have been used empirically for mitochondrial dysfunction, when present. An oral formulation of cholesterol dissolved in soy oil may normalize the cholesterol level but without obvious clinical benefit. One critically ill patient improved after receiving IV pantothenic acid, but many others showed no improvement. Future clinical trials may involve dietary supplementation with specific lipids.

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Barth Syndrome Family Support Network Evolves into The Barth Syndrome Foundation, Inc.

Our group, formally known as The Barth Syndrome Family Support Network, is now known as "The Barth Syndrome Foundation, Inc."

Upon unanimous vote of the family members in attendance at the Barth Syndrome Family Support Network Family/Scientific Conference which was held in Baltimore in June 2000, Anna Dunn, Sue Wilkins, Shelley Bowen, Kate McCurdy and Steve McCurdy embarked on our collective mission to transform our group into an official not-for-profit (NFP) organization. We were advised by not-for-profit attorneys, who had volunteered to help us on a *pro bono* basis to accomplish our goals, that we should change the name of the group. Statements made supporting this decision were the following:

- The IRS does not look favorably at granting NFP public charity status to groups which appear to support a limited population.
- An organization must prove ability to assist the public through their programs.
- A NFP public charity cannot support any one group or member through financial contribution.

In order to move forward with the objective of becoming a public charitable organization and to defend the application for this status, our name was changed.

However, we on the Board of Directors actually think it was a very good idea, not just because it made our case easier to present to the authorities, but also because the name change actually reflects a very important evolution of our group. We now are at the stage of "thinking big". Always with the boys and their well-being at the forefront of our minds, we now can consider not just helping each other by sharing our experiences and knowledge (though that will **ALWAYS** be **SO VERY** important), but also by potentially raising money and funding research grants of our own and encouraging wonderful and talented researchers to focus their attention (or at least some of it) specifically on Barth syndrome. We can form alliances with other organizations, such as the Genetic Alliance or the National Organization of Rare Diseases, to work together and to further our cause. We can create and get support to pay for an awareness program for pediatric physicians, possibly including mailings, articles and presentations. There is much we can do, all to help the boys we have and others out there who are not yet diagnosed, but we have to expand our horizons.

Steve McCurdy is a member of the Board and has taken on responsibility for long-term planning for our foundation. Recently, this has meant shepherding the legal incorporation of our foundation and obtaining 501(c)(3) tax-exempt status (which makes us a non-taxable and tax deductible charity) under US Internal Revenue Services rules. Although the BSF has been in existence for less than a year, I think you will be impressed by our progress so far, and by our plans for the future. Our vision is *a world in which not one more child will suffer or perish from this condition*. On September 8, 2000, our foundation was formally incorporated as the Barth Syndrome Foundation, Inc. On January 8, 2001, we received notification from the IRS that our application for 501(c)(3) status as a public charity was approved. We are now registered in six states wherein we can legally solicit funding and are prepared to register in any others where it may prove appropriate.

Salutation from Professor Peter G. Barth

A moment of pride...

This is a memorable event. First of all, it marks a row of memorable events, that all happened in the last two years. If there is anything that strikes me at this moment, then it is to see how sheer despair about the misery of so

"It is important also to present a platform to people with a common interest within the community, and this is exactly what the Family group under an excellent leadership has achieved."

many children, all boys with the same kind of profound illness, can be turned into a positive sense of a shared destiny and how this sense is being set to work.



Prof. Peter G. Barth

Many times I was told by parents how lonely one can feel when your child has a disease that nobody seems to know. I must admit that it took me years to discover the depth of this feeling, and I only realized it fully when I found myself in the midst of that wonderful first meeting of the organization in Baltimore in June 2000 and saw this gathering of parents, sibs and affected boys, happy with the occasion to share some of their common experiences – but also some of their plight - with others.

Sharing one's misfortune with others however, I am sure, is a good thing, but certainly it is not the only aim of this organization. It is important also to present a platform to people with a common interest within the community, and this is exactly what the Family group under an excellent leadership has achieved. The Newsletter, in addition to the website will help to further the aims of the Family group, which has matured and became the Foundation.

For one thing, I am sure that the Newsletter has its start at the right time. In just one decade the disease now generally called Barth syndrome was "mapped" on the X-chromosome, then the gene was identified and last year the cardiomyopathy defect was discovered. Indeed, we know much, much more than just a few years ago...but we are still far from our goal: to get more specifically aimed and better therapies. This should be a joint goal for the families and the scientists. I expect that the Newsletter will help to bring this about in many ways. Foremost, it will help to make others whose interest is vital to us aware of this rare disease.

I regard it as an honour to write these introductory words, and I take this opportunity to congratulate all those who have done so much to realize this great news.

Prof. Peter G. Barth,
Department of Pediatric Neurology
Emma Children's Hospital/ AMC
University of Amsterdam

Envisioning the Future

In early April of this year, members of the Barth Syndrome Foundation Board of Directors consisting of Shelley Bowen, Anna Dunn, Lynda Sedefian, and Kate and Steve McCurdy (along with Sue Wilkins checking progress and contributing by phone), met in Anna's kitchen to create a long-term strategic plan for the foundation. In preparation for this meeting we researched other successful foundations to determine what characteristics set them apart from others. For three days, we talked and wrote and covered Anna's walls with large sheets of paper filled with ideas, hopes and dreams.

We imagined a world where all pediatricians were familiar with Barth syndrome and could easily recognize and diagnose the disorder, and when once diagnosed, effective treatments would be easily and inexpensively available.

We envisioned the day when families did not have to struggle to find knowledgeable physicians or information on Barth syndrome or how to care for their affected boys. We also envisioned Barth families connecting with a loving and supportive community who would care for them, alienating the isolation, frustration and worry that is so much a part of coping with a rare disorder, by just a phone call or an Internet chat or a visit to an informative website filled with relevant information.

We yearned for the day when Barth syndrome was recognized as an important subject of medical research, and labs and researchers around the world devoted their time, energy and resources to understanding its genetic causes and developing effective treatments... and ultimately a cure!

We dreamed a lot in Anna's kitchen. The dreams of hopeful parents we all knew, indeed, our own dreams. And from these dreams we cooked up a plan.

Like any good plan, it had to start with a vision. Our vision is: *"A world in which not one more child will suffer or perish from Barth syndrome."*

The foundation had to have a clear mission that everyone could understand and support... *"To guide the search for a cure, to educate and support physicians and to create a caring community for affected families."*

But it couldn't stop there. It had to have a set of realistic, accomplishable goals, which would move us closer and closer to our ultimate vision. There would be no point in dreaming dreams with no hope of achieving them. We needed to be able to make progress toward those dreams, and be able to measure our progress so that we would always know that we were on track, moving ahead and not wasting our efforts and precious resources.

It had to be a plan that all of our families could easily understand and support. It had to be a plan that could convince foundations and individuals alike to contribute money. And it had to be a plan that would inspire our families and growing list of friends to lend their personal support, time and energy to. And so we eventually settled on five goals...

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.**
 - Research indicates that only 30% of undiagnosed Barth children will survive their first few years of life. An accurate diagnosis can increase that survival rate to 85-90%. Increasing awareness among physicians will save the lives of many children.
2. **To encourage, guide and fund additional research to improve diagnosis and treatment, and ultimately to develop a cure for Barth Syndrome**
 - The Barth gene has been identified and mapped. Sporadic research efforts are under way but no national or international funding or organization is focused on Barth syndrome, its underlying causes, or potential treatments or cures. Clearly, a more focused and organized research effort would ultimately help our families.
3. **To create a caring community that will offer each Barth family information, guidance and emotional support**
 - Before we found each other over the Internet and gathered in Baltimore, we all felt the same isolation and desperation, wondering if we were the only ones in the world who were struggling with this rare disease. No one should have to go through that, and we can make certain that no one does. We need to build a "community" to bridge the miles that separate us from each other.
4. **To build and sustain a broad base of concerned contributors who will provide the funds we need to accomplish our mission and goals**
 - Building a community, increasing awareness and especially funding research will require far more resources than our few families can ever muster alone. We will need to enlist the help of many other sympathetic individuals and organizations if we are to raise the money needed to achieve our goals and our vision.
5. **To create, inspire and make effective use of an organization of volunteers dedicated to reaching our vision... A world in which not one more child will suffer or perish from Barth Syndrome**
 - It is clear that a few people, no matter how determined, can never accomplish all that we envision. We will need to inspire more volunteers to help us reach our goals... and we will need to build an organization where they all feel welcome and encouraged to contribute their time, energy, enthusiasm, and skills to our cause.

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Envisioning the Future

(cont'd from page 5)

No small task! Now that we had dreamed our dreams, could we make them come true? Could we translate them into reality? That would ultimately depend on our ability to develop and implement an action plan for each of these goals that would take them from lofty goals on a piece of paper to a reality.

Fortunately, we had all done our homework. The National Organization of Rare Disorders has selected the top 20 foundations from among the thousands which are members. We researched and in several cases met with these foundations to learn from them what made them successful and to copy their best practices. We came with all the motivation that comes with having a child affected with Barth syndrome. Finally, we had some help from an outside consultant with experience in the non-profit world to add a strong dose of realism to our planning efforts.

What follows is a summary of the major objectives, which we developed for each of the goals listed above. Each of these objectives, in turn is supported by a more detailed set of action steps, which are not shown here, but which we would be happy to share with anyone who is interested!

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. Create an "awareness campaign" for the pediatric medical community (focused initially in the US and Canada)
 - B. Become a clearinghouse for scientific and clinical information on Barth Syndrome to assist physicians, families and researchers
 - C. Create a Medical Advisory Board (MAB) to provide advice and support for BSF's awareness campaign and to help physicians gain access to any specialized ancillary support services
2. **TO ENCOURAGE, GUIDE AND FUND ADDITIONAL RESEARCH TO IMPROVE DIAGNOSIS AND TREATMENT, AND ULTIMATELY TO DEVELOP A CURE FOR BARTH SYNDROME**
 - A. Create a Medical Advisory Board (MAB)
 - B. Identify strategy and parameters governing research grants to be offered by BSF
 - C. Expand the Barth Registry and Data
 - D. Stimulate interest in Barth syndrome research
3. **TO CREATE A CARING COMMUNITY THAT WILL OFFER EACH BARTH FAMILY INFORMATION, GUIDANCE AND EMOTIONAL SUPPORT**
 - A. Create and maintain information for families
 - B. Create opportunities for Barth families and boys to connect and communicate with each other
4. **TO BUILD AND SUSTAIN A BROAD BASE OF CONCERNED CONTRIBUTORS WHO WILL PROVIDE THE FUNDS WE NEED TO ACCOMPLISH OUR MISSION AND GOALS**
 - A. Raise sufficient funds annually to support the Barth Syndrome Foundation mission and goals
 - B. Validate and implement the fund raising program plans (\$785,000 in five years)
5. **TO CREATE, INSPIRE AND MAKE EFFECTIVE USE OF AN ORGANIZATION OF VOLUNTEERS DEDICATED TO REACHING OUR VISION... THAT NOT ONE MORE CHILD WILL SUFFER OR PERISH FROM BARTH SYNDROME**
 - A. Insure that the Foundation is always properly organized and staffed
 - B. Insure that the organization is clearly focused and guided by a clear set of operating principles
 - C. Insure that appropriate controls are maintained at all times

Our Goals are intended to address a period of five years, and will be reviewed and updated each year. Our annual plan will lay out the objectives and action steps we hope to complete during the following twelve months, and our annual report will detail our accomplishments during the previous year and our financial condition at year end. This document is intended to be a living, actionable guide to keep the Barth Syndrome Foundation firmly focused and to serve as a vehicle to communicate our progress toward our vision to our various constituencies: our families, our contributors, the medical and scientific communities and our volunteers.

We are now making excellent progress against many of these goals... but would love your help. If you feel you can help in any of these key areas, please contact anyone on the board and volunteer. We would like to hear from you!

DISCLAIMER AND PRIVACY POLICY STATEMENT

The Barth Syndrome Foundation Inc., (BSF) while designing and maintaining a website on the Internet that provides educational information to those in need, does not intend to provide medical advice. Also, BSF highly respects the privacy of each and every individual affiliated with our organization. Accordingly, BSF sought legal advice to compose the following Disclaimer and Privacy Policy Statement outlining BSF's role in complying with said issues.

Disclaimer

The Barth Syndrome Foundation, Inc. (BSF) website and newsletter are designed for educational purposes only and are not intended to serve as medical advice. The information provided on this site should not be used for diagnosing or treating a health problem or disease. It is not a substitute for professional care. If you suspect you or your children may have Barth syndrome you should consult your health care provider.

Web-Site Privacy Policy

The Barth Syndrome Foundation, Inc. (BSF) is committed to protecting your privacy. We have summarized our policies and procedures below. Your privacy is important to us. To better protect your privacy, we provide this notice explaining our online information practices and choices you can make about the way your information is collected and used. To make this notice easy to locate, we make it available as a link on our home page, and at every point where personal information may be requested. We define personal information to include: name, e-mail name, web address, physical address, phone number, survey responses, registry information and the content of your correspondence with us.

Disclosure

When you contact us, your email address, web name and other information included in your email may be forwarded through the site to the appropriate person(s) within The Barth Syndrome Foundation, Inc. (BSF) for a response. Copies of your correspondence may be retained at the BSF home office and by the responding individual. The Barth Syndrome Foundation, Inc. will establish and maintain lists of our members including families, physicians, scientists, contributors and others interested in Barth Syndrome to allow us to communicate with you. These lists will not be sold, rented or given to any third party for commercial use, nor do we currently expect to release these lists to any third party for any reason. However, if the Barth Syndrome Foundation sees a need or a possible benefit to you of making your personal information available to a third party, we will not do so without first identifying the recipient and providing a full explanation to you, and receiving your permission to do so in advance of each instance.

Name and Address

We use some of the information included in your e-mail message (e.g., your web name and address including your Internet service provider) to track usage of our web site and improve its value to you. We compile statistics that show the daily number of visitors to our site, the daily requests for particular files on the site, the countries that requests are coming from, and browser and operating systems used. These statistics are used internally to help us improve the content and scope of the site.

These statistics contain no personal information and cannot be used to gather such information. We may use this information to communicate with you, but will not distribute this information outside of BSF or use it for any other purpose without your permission.

Other Sites

The Barth Syndrome Foundation, Inc. (BSF) links to other sites that may be of interest to those who use our site. The BSF is not responsible for the privacy practices or the content of such Web sites.

Confidentiality

The Barth Syndrome Foundation, Inc. (BSF) is firmly committed to maintaining the confidentiality of your personal information in all of our activities and programs.

Every reasonable effort will be made to maintain the security of all personal information in our possession. Access to personal information will be limited to BSF Board members and officers and to individuals who have a need to use such information in the course of their work on approved projects for and overseen by the Barth Syndrome Foundation and who have been instructed in the confidentiality requirements of the organization.

WORKING TOGETHER WITH OTHER ORGANIZATIONS TOWARD A COMMON GOAL

The Barth Syndrome Foundation, Inc. (BSF) in an attempt to better advocate for our families, has fostered relationships with other organizations that are committed to serving the needs of families and individuals exhibiting similar components to those found with Barth syndrome. Below please find examples of such relationships that have been established:

- Genetic Alliance (<http://www.geneticalliance.org/>), knowing that Barth syndrome is an x-linked genetic disorder.
- For the first time ever the National Organization for Rare Disorders (NORD) (<http://www.rarediseases.org>) has recognized Barth syndrome as a rare disorder. BSF approached Gerald F. Cox, M.D., Ph.D., F.A.C.M.G., Medical Director, Department of Clinical Research, Genzyme Corporation, Cambridge, MA 02139; Assistant in Medicine, Division of Genetics, Children's Hospital, Boston, MA 02115, who agreed to compose an article detailing Barth syndrome which will be published in The NORD Guide to Rare Diseases.
- Barth syndrome is listed with the Birth Disorder Information Registry (<http://www.bdid.com/defectba.htm>)
- BSF has also formed an alliance with the United Mitochondrial Disease Foundation (UMDF) (<http://www.umdf.org/>)
- For the first time ever we have had the term Barth syndrome included as a potential cause of cardiomyopathy by the American Heart Association
- Prior to BSF becoming our own entity, the MAGIC Foundation (Major Aspects of Growth In Children for Children's Growth and Related Adult Disorders) (<http://www.magicfoundation.org/>) kindly took us under their umbrella organization enabling us to conduct business as a not-for-profit organization before we received such approval of our own

By enlisting other organizations, we may be able to combine resources in a common cause. Combining forces with similar disorders will also enable us to draw a greater audience. We are continuing to work on more alliances and will keep you posted on accomplishments as they unfold.

BSF's contributions to research have been cited to defend a research project submitted to the American Heart Association for funding by Dr. Michael Schlame. Additionally, BSF has written a letter to the American Heart Association in support of this research on behalf of Dr. Schlame.

NETWORKING TOWARD A SOLUTION

As someone who is affected by a rare disorder we can all understand the importance of being able to contact another expert or family member. Not so long ago the effective use of this group was demonstrated by one of our mothers, Karen Gordon. Karen was interested in knowing experiences with chicken pox. A mother of healthy children and a mother of a child with Barth syndrome, Karen realized there could be extenuating circumstances she should consider. She put our network to the test. Karen posted specific questions about this issue and received responses back almost immediately. Karen later wrote a summary of her responses to the group.

This encounter led our group to consider a way for our BSF members to interact immediately with other members and experts alike. We began to search for programs that could accomplish this goal and maintain the privacy of our members. This tool is called a listserv. A listserv is like a cyber file cabinet with specific discussions and chronological order in each discussion. The ability to archive data is very important. For example, a new family member will not have been a part of Karen's exercise; however, at some point in the future the parent will most likely need to know the information gathered through this poll. Wouldn't it be nice to have the access to this information in the future? The listserv offers that ability. In addition, the discussion, such as the one Karen started, would always be available for future comments. As time goes on a new family or even Karen may learn something more about her initial topic. They may go to that folder and make comments about that topic at any point in the future. There are many listservs available but we had specific criteria we wanted this tool to meet.

Privacy for our group: Your information remains private and not available to any commercial organization. The program we now use is through a grant with a specific purpose to service child and maternal health issues. This listserv is moderated by a BSF member. Any inappropriate discussion will not be posted. Members must sign up to participate in this group thereby eliminating the risk of intrusion. Commercial discussion groups such as MSN or Yahoo sell your information to other cyber-groups which opens the gates to junk e-mail. This group does not.

Retrievable discussions to refer to in the future: Karen's thoughtful questions and follow-up distributed the need for information and reference. With this program our group will be able to search previous discussions or threads in the future. New BSF members will be able to access this information to learn more about small but very important details regarding those affected by this disorder.

If you are interested in participating in this listserv go to <http://mchenet.ichp.edu/> to learn more. If you have any questions please do not hesitate to contact Shelley Bowen at sbowen@barthsyndrome.org

Survey 2001

I started this survey a few years ago out of curiosity about my boy. The replies were intriguing. I found that not all boys were the same but in many unusual ways they were similar. We have now found these data to be useful tools in answering questions posed by researchers and physicians. As a group, we have all contributed to science by taking the time to fill these surveys out. I know they are long, but again they are useful.

Because the previous two surveys were so cumbersome and difficult to compile, the Barth Syndrome Foundation, Inc. (BSF) Board has considered options to this dilemma. Mark Dunn has obtained a donated program to enable us to compile data over the Internet. It will offer instant results, but we are working out a few very important details to make these data useful to science. We are hoping to have this software posted to a private website allowing only affected families the opportunity to view and fill out this form. As soon as these details have been worked through we will let you know where you can go to fill it out **and ask that you take the time to do so**. It has been proven effective to assist medical community in research. We have polled various physicians and researchers about information they feel would be useful to know. In addition, we would like for you to respond in kind. Please respond directly to Mark Dunn with the following:

What questions would you like to see asked in the Barth Syndrome Survey 2001? You need only compile about five (5) questions but obviously if you have more these will be considered. Please submit your questions to: mdunn@barthsyndrome.org.

Shelley Bowen, President, BSF

BARTH SYNDROME GAINS MORE ATTENTION IN SCIENTIFIC ARENAS

As Barth syndrome gains more attention within the scientific community, research is accelerating. In 2000, for example, an underlying biochemical defect involved in Barth syndrome was first identified; and in 2001, steps were taken toward the development of a new, simple diagnostic test for the condition. BSF and several families provided direct support for several of these studies by organizing to provide tissue and blood samples for research.

The Barth Syndrome Foundation and several scientists are making efforts to engage the interests (and funding) of the National Institutes of Health. Kate McCurdy is in the initial stages of creating a Scientific and Medical Advisory Board (SMAB), as an adjunct to the BSF. The SMAB will consist of a group of internationally prominent scientists and clinicians with specialties in the areas most critical to Barth syndrome research. They will help advise the BSF Board on all scientific and medical matters and assist with the educational, research and awareness activities of the organization. We hope they will also join us at our bi-annual conference to answer our questions and brief us on the latest advances affecting us.

United we stand...

Although The Barth Syndrome Foundation, Inc. is a USA-based not-for-profit organization which has been established to support the needs of those families and affected individuals of this disorder, we are also committed to broadening our outreach efforts to assist all families and individuals affected by this disorder worldwide. Thanks to Joke van



Loo, a Family member located in Netherlands, Europe, a Dutch website about Barth Syndrome is on the Internet. You can find it at <http://www.stofwisselingsziekten.nl>. Click on VKS-logo (Vereniging voor Kinderen met Stofwisselingsziekten) and then on the link [Ziekten] on the left and in the list with all kind of metabolic diseases "Barth Syndroom". At the top of the page about Barth Syndroom are 4 links:

- [\[kenmerken\]](#) (Clinical Features)
- [\[diagnose en behandeling\]](#) (Diagnosis and treatment)
- [\[nuttige links\]](#) (with a link to www.barthsyndrome.org)
- [\[andere ziekten\]](#) (back to list of metabolic diseases).

Dutch families can get support from the Vereniging voor Kinderen met Stofwisselingsziekten. Joke van Loo will be the contact person for the Dutch Barth Syndrome Group and may help Dutch families with translation of American pages about Barth syndrome, with explanation of scientific articles about Barth syndrome and give addresses and telephone numbers of helpful organizations and social regulations as financial support in the Netherlands.

This is a terrific example of accomplishment by more of our determined volunteers! Joke, we congratulate you on your efforts to assist BSF in our outreach endeavors and encourage more of our volunteers to do the same.

DEVELOPMENT EFFORT OFF TO A QUICK START

When the Barth Syndrome Foundation (BSF) board got together in Anna Dunn's kitchen to create our five-year strategic plan (see the related article on the strategic plan), we all became very excited by the kind of impact we realized we could have on our families, our boys and future generations. We began to map out plans for family support (website, newsletters, medical resources, educational brochures, 2002 family conference, etc.) for increased awareness among pediatric physicians so that they can recognize Barth syndrome, save lives and treat our boys appropriately, and for medical and scientific research to help develop inexpensive diagnostics, treatments and cures. We also realized that all of these programs would require funding if we were to be successful. Our initial goal is to raise approximately \$785,000 over five years.

At first, this sounded like an impossible task. Particularly since we have identified only about 40 families in the U.S. and Canada so far, and every one of these families has its hands full simply taking care of itself. But then we did our homework. We researched a number of other successful foundations focused on rare disorders and we consulted with an experienced fund-raiser – Richard Kearns, who works with a number of prominent New York area hospitals. With Richard's help we developed a plan, which we have already begun to put into action.

Individual Contributions

The first step in our fund raising plan is our direct appeal to our families and friends. In the U.S., our status as a tax-free foundation, and our ability to attract tax-deductible contributions depends on our receiving at least 30% of our financial support from smaller individual contributions (including those made through fundraising events). Large contributions, even though important to us, do not count toward this requirement. The IRS wants to be assured that our foundation seeks to serve, and enjoys support from a broad public base.

Likewise, when we go to foundations, businesses and potential donors of major gifts, it is important that we be able to make a strong case to them to support our foundation (BSF) over many other possible charitable causes. One of the most important statements we would like to be able to make is that we have the support of 100% of our families, and have received donations from a large number of contributors.

For these reasons, while smaller contributions from individuals are not expected to be the largest source of funds by any means, it may be the most important. Those of you who are family members have recently received a letter from Steve McCurdy, which described our progress in setting up the foundation since our last meeting in Baltimore. In the letter, Steve asked you to consider making a donation in any amount you feel is appropriate – whether \$1 or \$100. We know that our families already devote all their efforts, time and money, to the care of their children and are the real “angels”. But we felt we had to go to you first, before we spread our development effort further.

Its early, but we have already received a number of donations from the U.S. and abroad indicating a high degree of support from our most important constituency! Our next step is to broaden our appeal, with the help of our families and early supporters, to include friends and other members of our local communities.

We have developed an Informational Brochure to increase awareness and encourage contributors and we have made every effort to make giving to the Barth Syndrome Foundation, Inc as easy as possible. We now accept VISA, MasterCard and American Express. We can accept donations on the Internet through our website, www.barthsyndrome.org, and of course we can accept checks sent to Sue Wilkins, Treasurer, c/o Barth Syndrome Foundation, Inc. at P.O. Box 23173, Lincoln, NE 68542-3173.

Major Gifts and Grants

Most of our funding over the next five years is expected to come from grants and major gifts. We have been incredibly fortunate to have the assistance of Rosemary and Mary Baffa, who contacted and helped us convince a major foundation to give us a single grant of \$100,000 specifically for research. That money has now been received, and under the guidance of the BSF Scientific and Medical Advisory Board which Kate McCurdy is forming, will soon be put to good use in selected research labs. Thanks to Rosemary and Mary, we have gained valuable credibility as we contact other foundations in search of additional funding. We have also been blessed with a major gift – a founding donation - from Paula Varner, Sue Wilkins' mother, which has enabled us to accomplish all that we have so far. Finally, we have also received a challenge grant from an anonymous donor who will double match all contributions received from families of boys affected by Barth syndrome, up to \$10,000 in total. This means that every \$1 contributed by a family member will turn into \$3 when matched by this challenge grant. This is the kind of support that is usually only given to larger, more established foundations and we are very lucky to have attracted this kind of support so early!

Fund Raising Events

Another important source of fundraising should come from a series of events such as the Walk-a-thon that Lynda Sedefian ran, which raised over \$15,000 in one day. We hope that more of our families, supporters and friends will volunteer to organize events such as Lynda's. Each of us can find ways to contribute in our own way. Cherie Schrader, the sister of Carol Cook, has volunteered to raise money by finding sponsors to support her as she runs in the Chicago marathon in October. As an accomplished runner who earns her living in the restaurant business, Cherie is asking friends to contribute “by the mile”, and small businesses, particularly restaurants with whom she has a connection, to “buy a body part”! She will advertise their support for BSF by painting the restaurant's name on various parts of her body. If you happen to be in Chicago on October 17th, be sure and join Cherie's family and friends to cheer her on and to support the Barth Syndrome Foundation!

(cont'd on page 11)

DEVELOPMENT EFFORT OFF TO A QUICK START

(cont'd from page 10)

Lynda Sedefian and Shelley Bowen have developed a simple, cook-book approach to creating a successful fundraiser. It provides step-by-step guidance to help you organize your event. You will find that many organizations such as the Kiwanis Club, local businesses, as well as larger companies with local stores or offices such as Wal-Mart are happy to help. Please let us know before you get started, as we may need to register the Barth Syndrome Foundation in your state or province before you begin fundraising. Either Lynda Sedefian or Steve McCurdy would be happy to help you get organized – you can contact us through the BSF web site at www.barthysyndrome.org!

And finally, we are also looking for additional volunteers to help us launch and support fundraising events, or provide us with leads to foundations and major contributors. If you would like to help, please contact Steve McCurdy or Lynda Sedefian who will be happy to hear from you!

There is still so much to do, and our family support, physician awareness and research projects all depend on our ability to raise funds to support them. These are very important goals, and we need your continued support to help us reach our vision, that **“Not one more child should suffer or perish from Barth syndrome”!**

Submitted by Steve McCurdy

\$100,000 GRANT RECEIVED

Perhaps the most exciting news is that through the extraordinary efforts of Rosemary Baffa and her family, the Barth Syndrome Foundation has been given a \$100,000 grant for research and to encourage awareness, by a private foundation, which prefers to remain anonymous. Although we cannot name our benefactor, we can certainly extend our thanks to Rosemary and her family. This grant, so early in the life of our foundation, gives us immediate credibility in further development efforts and within the research community. It is the seed money from which we hope a might research oak will grow!

Walk-a-thon A Huge Success

(cont'd from page 1)

This event, sponsored by New Scotland Kiwanis and Broadway Marketing, brought together many folks within Sedefian's community. "I would particularly like to thank the students of Voorheesville Elementary School, Clay R. Bouton Jr./Sr. High School, and the Greenville Central School District for their participation in this event. It was heartwarming to see these students truly wanting to make a difference and become a huge part of this event." With the assistance of other local community organizations such as Wal-Mart and Sam's Club, matching funds were also obtained.

Public awareness was also a very important part of this fundraiser, with the distribution of well over 1,000 educational brochures throughout the community. As a result, a connection was made with a family residing in Sedefian's community who is in the process of undergoing tests to determine whether that family has a son with Barth syndrome.

"The community was so very supportive, as well as friends and family who worked hand in hand to make this day such a memorable one. I must admit that going into this I had no idea what the outcome would be other than knowing for certain that I would, at the very least, have distributed BSF's informational brochure within my community, which in and of itself is a very important part of this foundation. I look forward to doing it again next year."

Submitted by Lynda Sedefian



Lynda and Derek Sedefian, acting on behalf of BSF, were presented with matching funds from Shay Green, Store Manager, Albany Wal-Mart.

Shay Green, Store Manager of the Albany Wal-Mart presents BSF with a \$500 check from Wal-Mart's Community Matching Grant Program, the largest program funded by the Wal-Mart Foundation. Raffle tickets were sold in front of both Wal-Mart and Sam's Club prior to the walkathon, and the profit from the sale of these tickets was matched by both Wal-Mart and Sam's Club.

Calling all Volunteers...Should you wish to assist BSF in raising funds by organizing an event such as a walkathon, a golf outing, a dinner along with a silent auction, a sports night or any other event, The BSF can help you prepare and provide a number of useful tools such as a video which can be shown at the event, informational brochures and "how to" guidelines. However, since each state's laws are different, we ask that you get in touch with Steve McCurdy (smccurdy@barthysyndrome.org) or Lynda Sedefian (lsedefian@barthysyndrome.org) if you are thinking of sponsoring an event or doing any organized fundraising. We can help you (and us!) stay in compliance with your state's laws governing charities and fundraising.

A Summary of Research into a Possible “Cognitive Phenotype” in Children with Barth Syndrome

Based on research that was conducted by Michèle .M. Mazzocco, Department of Psychiatry and Behavioral Sciences, Johns Hopkins University School of Medicine, Math Skills Development Project, Kennedy Krieger Institute, Baltimore, Maryland and Dr. Richard Kelley, Department of Pediatrics, Johns Hopkins University School of Medicine, Department of Metabolism, Kennedy Krieger Institute, Baltimore, Maryland, a report concerning a “cognitive phenotype” in children with Barth Syndrome (Mazzocco, Kelley, 2001) recently appeared in the *American Journal of Medical Genetics*. The report is undoubtedly of interest to the readers of this newsletter, and for this reason it is summarized here. The summary is presented in the context of a discussion of why we should ask the question: “Do boys with Barth syndrome have a “cognitive phenotype?” The recently published report is an initial, preliminary study that was designed to address this question, but also to address the importance of the question itself.

What is a cognitive phenotype?

When scientists explore evidence for a cognitive phenotype, their primary question concerns whether children with a specific condition have specific characteristics that will affect their cognitive development and, in turn, their academic performance. Just as a physical phenotype may include expression of specific physical traits (such as short stature), a cognitive phenotype may include characteristic features of intellectual development, academic achievement, and learning. A cognitive phenotype may be “specific” in terms of predisposing children with a particular condition to have more --or less-- difficulty in a particular area of functioning (such as reading, math, language comprehension, attention skills, or spatial problem solving). However, even when a specific area of functioning is identified, that identified cognitive strength or weakness is rarely observed *only* in persons who have the condition in question. That is, *if* boys with Barth syndrome have a specific cognitive phenotype, that phenotype will include cognitive strengths and/or weaknesses that are observed with greater frequency among boys with Barth syndrome, relative to boys in the general population. However, those strengths or weaknesses may also be observed among some children who do not have Barth syndrome; and not *all* boys with Barth syndrome will demonstrate those strengths and weaknesses. Whether this “cognitive profile” includes learning disability depends in part on the definition of a

learning disability; a discussion of this definition is beyond the scope of this brief article. Regardless of how the learning profile is labeled, it is important to study whether children with Barth syndrome have a cognitive phenotype.

Why study a cognitive phenotype?

If there is indeed a cognitive phenotype of Barth syndrome, information about the phenotype can guide parents and practitioners in early identification and intervention. Early intervention may affect the developmental course of certain skills by allowing or encouraging a child to exercise an area of weakness, by exposing a child to skills known to be related to areas of potential difficulties, or by enhancing compensatory skills through the child’s strengths. For example, if a child is at risk for reading disability, one can draw upon the vast research on the building blocks of reading and provide the child with exposure to activities that strengthen the very weaknesses that may lead to a reading difficulty. Identification of strengths can also guide practitioners to facilitate appropriate compensatory mechanisms, or a route to deal with a specific weakness. For example, if a child consistently fails to remember which sign is the “more than” (>) or “less than” (<) sign in mathematics because the signs are *spatially* difficult to differentiate, teaching the child that the larger number is on the sign next to the larger opening may enable the child to learn the symbol in a less abstract manner. Identification of strengths also provides a foundation from which to plan activities for success experiences, because children with learning difficulties may experience frustration with repeated difficulty or failure.

The underlying reasons for all learning and cognitive skills are not known, and one way to identify the skills that underlie a cognitive strength or weakness is through studies of what differentiates cognitive performance among children who are successful versus unsuccessful on a specific task. That is, we can learn more about how children with Barth syndrome achieve in school, and why learning in some areas may be harder or easier for them than for other children, by examining whether there is a cognitive phenotype in Barth syndrome.

How do we identify the cognitive phenotype of Barth syndrome?

There are many challenges to pursuing this type of work. One major challenge is that there are, in general, vast individual differences seen in child cognitive development.

Children inherit many traits that are expressions of genes unrelated to Barth syndrome, and children are exposed to environmental factors that may also affect their cognitive development. In part because of the interaction of these and other factors, some children with a specific syndrome may not demonstrate the cognitive phenotype associated with that syndrome; alternatively, a child with a specific syndrome may demonstrate difficulties (such as reading disability) that are unrelated to the syndrome but related to other genetic or environmental factors. The goal of cognitive phenotypes is to identify which cognitive characteristics are linked to Barth syndrome. This can only be done by studying a large group of children who have Barth syndrome – a group that is “representative” of the overall population children with Barth syndrome. It would be misleading to base a study on only those children whose parents and teachers report poor academic achievement, or on only those children for whom no such concerns exist. Thus examining cognitive performance among a large, representative group of children with Barth syndrome can inform us of whether there is a specific pattern of learning strengths and weaknesses in these children.

In the preliminary published report, the study was based on only five young boys with Barth syndrome, so it is far from conclusive. In addition to the small sample, the study was limited to an abbreviated psychoeducational test battery that was administered to each boy, rather than the more extensive battery of tests often used in cognitive phenotype research. Using data available from a large ongoing study, scores from 120 boys of similar age or grade level were used for a comparison group; these boys did not have Barth syndrome.

Comparisons between these two groups reflect some differences. Relative to boys in the comparison sample, boys with Barth syndrome had comparable performance on reading and pre-reading skills, and there was no suggestion of group differences on these skills. Although no statistically significant group differences emerged on the math measures, the majority (but not all) of the boys with Barth syndrome scored in the low average to below average

Barth Syndrome Foundation Participates in a Study Revealing New Insight into the Deficiency of Cardiolipin in Children with Barth Syndrome

About one year ago, on 12 September 2000 I decided to go over a pile of computer print-outs with biochemical data on muscle biopsies that our lab had obtained from Dr. Salvatore DiMauro (Columbia University, New York). It was the latest analytical run in a long series of experiments in which we had screened mitochondrial muscle diseases for involvement

"It looked unlike anything I had ever seen in muscle tissue. The dominant molecular form of cardiolipin was entirely missing but some abnormal cardiolipin molecules had accumulated in the tissue."

of a specific mitochondrial molecule - cardiolipin. I was not particularly excited since our measurements had not revealed anything interesting ever since we started this project about four years ago. However, on this day, one analysis immediately caught my eye. It looked unlike anything I had ever seen in muscle tissue. The dominant molecular form of cardiolipin was entirely missing but some abnormal cardiolipin molecules

had accumulated in the tissue. I called Dr. DiMauro's laboratory and was told that the patient from whom this biopsy was taken was diagnosed with Barth syndrome. I did not realize at the time that this would be the beginning of an engaging research project that would let me experience an unprecedented alliance between patients, their families, clinicians and researchers.

I have to admit, last year in September I had almost no knowledge of Barth syndrome. I certainly had not heard about it in medical school. So, I proceeded to do more reading and quickly became familiar with a number of authorities in the field, such as Dr. Peter Barth and Dr. Richard Kelley. I then began contacting them to find out what they thought about our new results and learned that they had already considered cardiolipin as a factor in the disease process. We all agreed that this line of research ought to be continued, but also realized how difficult it would be to obtain tissue samples from patients with confirmed Barth syndrome. It was Dr. Kelley who mentioned to me the existence of a family support group with a web site on the internet. He thought that this might be an avenue to recruit patients for our study and to make contacts with other interested physicians. This

is how I learned about The Barth Syndrome Foundation and decided to approach Shelley Bowen, its president.

I was surprised about how quickly things evolved from there. Shelley Bowen instantly circulated our request to recruit more patients among members of the foundation and within a couple of days I was receiving messages from various families who offered their support. At this time, I began to realize the devastating effects of this disease on patients and their families. I was also very impressed with the immense determination of the Barth families in doing whatever possible to help their children.

Only one week after my initial contact with Shelley Bowen, I went to New Jersey, where I met for the first time a 7 year old patient with Barth syndrome and his mother. Up to then, the idea that cardiolipin might be involved in Barth syndrome was just a vague possibility. After all we had only examined a single muscle biopsy and therefore we still needed to verify the result in other cell types such as platelets. The patient and his mother agreed to donate a blood sample for the study. The sample was drawn and processed on the same day back in our laboratory in the Hospital for Special Surgery in New York. The result was striking: We found almost no cardiolipin in the platelets of our patient, which was in contrast to platelets from healthy children. From this point on I had no doubt that we had found a new lead in our research on Barth syndrome.

In the following months, we continued to rely on The Barth Syndrome Foundation to recruit patients and to establish contacts with colleagues in the US as well as abroad. The convenience to establish new professional relationships was an instrumental factor in the progress we made. The contacts that turned out to be most important were Dr. Jeffrey Towbin from Houston and Dr. Peter Vreken from Amsterdam. Both had been active in Barth syndrome research long before us and were ready to collaborate and to contribute important ideas to our project. It was a big shock to us when Peter Vreken suddenly passed away, only a few months after we had begun to hope for an active collaboration with him and his group.

When we finally decided to publish the results of our study, I again became painfully aware of the many deficits in my knowledge about Barth syndrome. And once more I turned to the foundation for help. Shelley

Bowen had compiled a comprehensive survey of symptoms and features of patients with Barth syndrome. The survey was based on a number of



Michael Schlame, M.D.

questions that were passed on to the parents of affected children. It covered a broad range of topics, such as symptoms, disease progression,

and treatment options. I found the survey to be an excellent resource. Not only did it contain a record of most of our study patients; but it also allowed me to compare their clinical symptoms to other Barth patients. In short, it gave me a much better perception of the disease.

Within the last year, strong evidence has accumulated that children with Barth syndrome have cardiolipin deficiency, and there is at least suggestive evidence that this deficiency is pertinent to the disease mechanism. We also have a new biochemical method to diagnose cardiolipin deficiency in blood samples, rather than muscle biopsies. This method will be useful to distinguish Barth syndrome from other forms of cardiac disease in children. This progress would have not been possible without the support of The Barth Syndrome Foundation. In addition to establishing contacts, recruiting patients and collecting data, the foundation has provided emotional support to me and my colleagues.

Submitted by,

Michael Schlame, M.D.
Department of Anesthesiology
Hospital for Special Surgery
Manhattan, New York

Barth Syndrome Believed to be the First Known Human Disease caused by a "Lipid Remodeling Defect"

Dear Friends,

Since we last spoke at the June 2000 Baltimore meeting, some exciting scientific developments have occurred that are of direct relevance to

"By organizing into an effective group, you have expanded the awareness of Barth syndrome around the world, helped raise funds for research, and educated many people about the disease, who otherwise, would have been ignorant. For these efforts, I salute your hard work and single-minded dedication to defeating this disease."

you. Before discussing the science, I would like to thank the Barth Syndrome Foundation for such long-term support and cooperation with the scientists and health-care providers across the globe. Your efforts have made all the difference in helping us grapple with this disease. Because you have worked so hard at answering questionnaires, donating biological samples for both diagnostic/research purposes, and sharing personal thoughts about symptoms, you have helped advance our understanding of the disease. Your continued cooperation and support will speed the search for viable treatments.



Troy Phipps, Doctoral Student

By organizing into an effective group, you have expanded the awareness of Barth Syndrome around the world, helped raise funds for research, and educated many people about the disease, who otherwise, would have been

ignorant. For these efforts, I salute your hard work and single-minded dedication to defeating this disease.

The Barth Syndrome Foundation is a terrific organization full of the most wonderful people that I have had the pleasure to know. What started as a research project turned into a love affair with your cause, because you always kept me in touch with the fact that Barth Syndrome is a human disease and that failure is NOT an option.

Science Update

In January 2001, Dr. Peter Vreken, whose lab solved the primary biochemical defect in Barth syndrome, passed away in Amsterdam, The Netherlands leaving behind a wife and children. Although we will miss him dearly, his work is already recognized as a major advance in our understanding of Barth syndrome and human biochemistry. The scientists who knew Dr. Vreken felt that he deserved to be honored for this important discovery, so the biochemical reactions that he and his research group discovered will be called the **Vreken Pathways** in future publications.

So what are the Vreken Pathways and how does it help us understand Barth Syndrome?

Dr. Vreken (and the others in his research group) discovered that Barth syndrome is due to a lack of certain types of fat-like chemicals (called lipids) that are found in all animals. Lipids are critical components of cellular membranes, and are very important because they give structure and order to the cell and its interior, and can also be used for energy production. The team found that cells from boys with BTHS syndrome had reduced amounts of certain forms of **CL** (cardiolipin) and **PG** (phosphatidylglycerol). Please

DO NOT attempt any dietary intervention without involved discussion with your pediatrician. Dietary supplementation could lead to extremely dangerous side effects!!!

Normally, cells can make CL and PG, but before being used, they must be chemically modified by different enzymes. The data led Dr. Vreken's group to conclude that the BTHS protein may be one of those lipid modifying enzymes (see Figure 1). The official term used to describe the biochemical defect is a "lipid remodeling disorder." What makes BTHS syndrome so special is that it is the first known human disease that is caused by a "lipid remodeling defect."

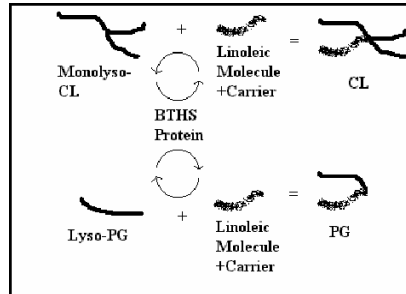


Figure 1. A simplified, hypothetical drawing of the proposed Vreken Pathways that leads to modified lipids. The BTHS proteins may help remodel both Lyso-CL and Lyso-PG into CL and PG. It is likely that the linoleic molecule is attached to a carrier molecule in the body.

How do the hypothetical Vreken Pathways help us understand the cardiomyopathy?

Our understanding of the biochemistry underlying Barth syndrome is still incomplete and will change over time, but I want to present the most important advance in our understanding based solely on published scientific literature. CL is found only in particular regions of some cells, called mitochondria. Mitochondria are extremely important structures inside cells, and most cells contain thousands of them. Mitochondria are protected from the rest of the cell's interior by two lipid membranes (an outer and inner layer). They produce almost all the energy for a cell from the food we eat and the oxygen we breathe, plus they perform a multitude of other life-sustaining biochemical functions. If the body cannot produce enough of the right kinds of CL and/or PG (which should normally go to the mitochondria), then the shape of the mitochondria may change, thus affecting the ability of the mitochondria to function normally (see Figure 2).

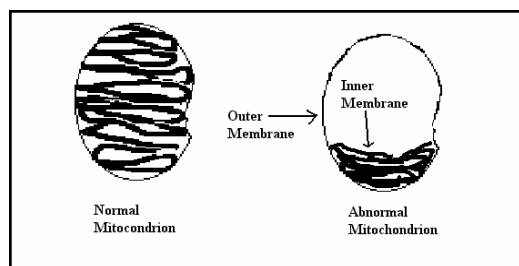


Figure 2. Drawing of normal and abnormal mitochondria. A common finding in BTHS syndrome is abnormal mitochondria as

depicted on the right. Abnormal mitochondria do not function as they should, and have stacked inner membranes.

All the functions of CL and PG are not yet known in humans. One established role for CL is in the normal functioning of a series of proteins, called the respiratory chain, that are located at the inner mitochondrial membrane and are critical for energy production. Our hearts and brains use the most energy, so it is very important for these tissues to have properly functioning mitochondria. The cardiomyopathy may be due to the body trying to

(cont'd on page 15)

Barth Syndrome Believed to be the First Known Human Disease caused by a “Lipid Remodeling Defect”

(cont'd from page 14)

compensate for a slightly reduced amount of energy and metabolic function from abnormally shaped mitochondria by laying down extra muscle layers. While this is a good short-term solution for keeping the heart strong, in the long-term, a thicker muscle can lead to less-efficient pumping.

The root problem in Barth syndrome may be a lack of the correctly remodeled lipids. Current drug treatments do not fix the primary defect, rather they help manage symptoms that are caused by a lack of these lipids. In the future, an ideal, effective treatment must re-establish the body's natural balance of lipids for the symptoms to hopefully reduce. However, CL cannot be eaten directly because it is broken down in our stomachs, and it cannot be injected into muscles, as it is probably **toxic**.

The CL type that seems to be reduced in BTHS cells is chemically modified with linoleic acid. By eating food enriched in linoleic acid, there might be an increase in available linoleic acid, which the BTHS cells might tag onto CL. **As stated previously in this article, please DO NOT attempt any dietary intervention without involved discussion with your pediatrician. Dietary supplementation could lead to extremely dangerous side effects!!!** This theoretically could increase the amount of the deficient lipid, which could then be used in mitochondria to stabilize the inner membrane and the respiratory chain. On the other hand, because children with BTHS may not be able to use the extra dietary linoleic acid, increasing the tissue levels of linoleic acid might interfere with the synthesis of other phospholipids that appear to be taking the place of CL in BTHS mitochondria, **thus leading to very dire health problems**. Although no one knows whether linoleic acid would have a **beneficial or harmful effect** in boys with BTHS syndrome, Dr. Vreken got us all thinking about the biochemistry of CL and how to turn this new knowledge into treatments that may safely and effectively treat BTHS syndrome.

While the future is bright for developing even better treatments, your group can facilitate this process by continuing to support all the professionals working on the disease, and by actively participating in the funding process that will pave the way for improving the quality of life in boys afflicted with Barth Syndrome.

Submitted by,

Troy Phipps, Doctoral Student
University of Southern California
Institute for Genetic Medicine
Department of Biochemistry
Los Angeles, CA.

A Summary of Research into a Possible “Cognitive Phenotype” in Children with Barth Syndrome

(cont'd on page 15)

(cont'd from page 12).....range on math achievement tests. Similarly, most of boys with Barth syndrome had below average scores on a visual-motor task, such as when copying shapes or figures. On visual tasks that did not involve motor planning such as copying, most (but not all) of the boys with Barth syndrome had more difficulty relative to the boys in the comparison group. Although these findings are based on a small sample size, the preliminary data presented in the published report are the first indication of a possible cognitive phenotype associated with Barth syndrome. More than anything else, the report provides a strong rationale for the continued study of a cognitive phenotype in boys with Barth syndrome. Yet it does not paint a complete picture of how the child with Barth syndrome learns.

It is important to pursue this research because of its implication for these children's future and their quality of life, in terms of academic achievement and cognitive functioning. The frequent reference to “normal cognitive function” in published reports of Barth syndrome suggest that, if there is a Barth syndrome cognitive phenotype, it does not include mental retardation. This is certainly the case in the preliminary study, where all five boys who enrolled had average intelligence, based on a verbal intelligence estimate. There are disorders with a relatively specific cognitive phenotype in the absence of mental retardation, such as Turner syndrome [Rovet, 1993], and approximately half of females with fragile X syndrome [Rousseau et al., 1994]. It is possible – but not conclusive -- that a mild yet specific cognitive phenotype is associated with Barth syndrome.

Research on the Barth syndrome cognitive phenotype will help to inform and guide the parents, teachers, and clinicians of a child with Barth syndrome. Knowing what potential features to look for may lead to earlier identification, and thus to earlier intervention. This can have a greater impact on academic success than later intervention, and may lead to specific educational guidelines based on what is known about learning disorders with similar learning profiles. Before this can be done, we need to replicate the present study with a larger sample, and with a more comprehensible test battery. Also needed are more in-depth assessments of the areas in which deficits are implicated, such as the assessment of different components of visual spatial skills. It will be important to study the development of these features over time, particularly as a function of treatment outcomes, genotype variants, metabolic variables, and the development of other markers of the disorder. In the meantime, the educational needs present in boys with Barth syndrome should be addressed as they would for any child who shows difficulties in specific academic skills, using resources available to help children with academic strengths and weaknesses.

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Submitted by Michèle M. Mazzocco, Department of Psychiatry and Behavioral Sciences, Johns Hopkins University School of Medicine, Math Skills Development

In the News.....

At the time of BSF's April walkathon, Lynda Sedefian contacted Sylvia Wood, staff writer for the Times Union in Albany, New York to encourage coverage of the event. Sylvia Wood took an interest in finding out more about Barth syndrome. As a result of this connection an article was written referencing Barth syndrome and was posted on Tuesday, June 5, 2001 in the Albany, New York Times Union, which states in its entirety:

RARE ILLNESS DIFFICULT TO DIAGNOSE By SYLVIA WOOD

The first clue that something was wrong with Lynda Sedefian's son, Eric, was his size.

"At the age of 1, we were concerned because he really wasn't growing," said Sedefian of Voorheesville.

But doctors found nothing wrong with the boy. Six months later, after a bout of bronchiolitis, Eric died. Sedefian had to wait another three years, until her third son almost died of heart failure, to get a diagnosis of the rare genetic illness that had affected the two boys: Barth Syndrome.

"Dr. Jeffrey Towbin, a professor of cardiology at Baylor College of Medicine and Texas Children's Hospital in Houston, believes progress is being made in educating the medical community about Barth Syndrome. He estimates the number of diagnosed cases of Barth Syndrome will increase significantly..."

"We never heard of it," she said. "At the time, there were only 20-some cases diagnosed worldwide. It was very difficult because there were no other families to connect with."

Yet, Sedefian considers her son, Derek, now 7, to be fortunate. Diagnosed when a newborn, he overcame the first obstacle that often stands in the way of survival for victims of this little-known illness that affects only males.

Without an accurate diagnosis, about 50 percent of all boys with the disorder will die, often before they are 2 years old.

"It's so rare, it's mentioned only as a footnote in most textbooks," said Dr. Richard Kelley, an assistant professor of pediatrics at Johns Hopkins University and an expert in metabolic disorders.

He estimates the incidence of Barth Syndrome at about one in 400,000 to 500,000 births, far less than one of the more common metabolic disorders, PKU, still considered rare with an incidence of about one in 15,000 births. The illness was only recognized in 1983, when Dutch physician Peter Barth studied a family in which all the male children died.

The most common symptom is cardiac failure. But boys with Barth Syndrome will often have other problems, including a condition known as neutropenia or extremely low white-blood-cell counts that make them more susceptible to bacterial infections.

Because the disorder also affects the body's ability to make cardi-olipin, a key nutrient needed for energy by the cells, boys with the disorder are often very small for their age.

For 14-year-old Michael Bowen of Perry, Fla., the hardest part of living with the disease is not being able to play competitive sports and being shorter than his friends and classmates. At 4-foot-2, he's

closer in size to grade-school children. "People tease me about my height," he said. "I just try to ignore them."

Bowen's mother, Shelly, has been on a mission to increase awareness of the illness since the death of her first son, Evan, in 1990, when he was 4 years old. "He was beautiful, articulate and bright," she said. "Both of my boys were misdiagnosed."

But Bowen was able to get a diagnosis for her son Michael, shortly after Evan's death. Since then, she's spent hundreds of hours on the Internet, trying to locate other families with the disorder and organizing the Barth Syndrome Foundation (<http://www.barthsyndrome.org>), which estimates that only 100 cases have been diagnosed worldwide.

"We have a lot of obstacles to overcome," she said.

"Without cardiologists knowing about the disorder, it's impossible to diagnose it. It's like describing the color red to someone who has never seen it."

"Dr. Jeffrey Towbin, a professor of cardiology at Baylor College of Medicine and Texas Children's Hospital in Houston, believes progress is being made in educating the medical community about Barth Syndrome. He estimates the number of diagnosed cases of Barth Syndrome will increase significantly over the next five years as more physicians recognize the disorder, which has only recently begun to receive more attention at national meetings and in medical journals.

"I'm quite sure it's underdiagnosed," Towbin said. "If you've never heard of the disease, you're not going to look, you're not going to find."

Physicians are learning that with proper treatment, boys affected by Barth Syndrome can improve over time, especially after the two rapid growth periods of infancy and puberty when energy demands on cells are at their highest. Eventually, some boys will even reach normal or close to normal height.

"Once the growth stops, the need for making cardi-olipin is reduced," Kelley said.

Although there is no cure, doctors have been able to improve the prognosis of boys with Barth Syndrome with new antibiotics to treat infections and with drugs to stabilize the heart disease. Some foods, including eggs, also seem helpful because they contain a nutrient that's similar to the missing cardi-olipin.

The goal of researchers is to figure out how the body makes cardi-olipin and to come up with a way to supplement the diet with the needed nutrient. "We're still doing a lot of footwork to understand the biochemistry," Kelley said.

Sedefian, who has organized local fund-raisers for the Barth Syndrome Foundation, is hoping the most difficult days are over for her son.

"The first couple of years are the hardest," she said. Derek's heart has recently been stable, and besides his small stature, he looks healthy.

"You would have no idea that he's gone through so much and that he lives day-to-day with this life-threatening disorder," she said.

"Reprinted with permission of the

The Power of Kindness.....

The Barth Syndrome Foundation, Inc. (BSF) would like to acknowledge and sincerely thank the following people for their contributions to our organization:

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In Memory of Peter Vreken, Ph.D.



The Barth Syndrome Foundation would like to express its sense of loss at the untimely death of Peter Vreken. His research at the University of Amsterdam in The Netherlands contributed much to important breakthroughs in the understanding of Barth syndrome.

His outstanding accomplishments, achieved during his short life, may potentially lead to a treatment and ultimately a cure for this rare and serious disorder, and for this we are extremely grateful. We would like to thank Dr. Vreken for both his direct contributions and his collaborative spirit and we offer his family our deepest sympathy. We will always remember Dr. Vreken as a dedicated researcher and a caring, generous human being.

The Barth Syndrome Foundation

If you would like to read the article written about the remodeling of cardiolipin as described by Peter Vreken et al request it from your child's attending physician and provide the information listed below to him or her. Many times the attending physician has access to a medical library through the institution or hospital of which he or she is affiliated and may be able to request this information in its complete format. If you are not able to gain access to this article in its complete format please refer to the BSF web site for a link to the abstract of this article.

Defective Remodeling of Cardiolipin and Phosphatidylglycerol in Barth Syndrome. Biochem Biophys Res Commun 2000 Dec 20;279(2):378-382

Vreken P, Valianpour F, Nijtmans LG, Grivell LA, Plecko B, Wanders RJ, Barth PG.

Department of Clinical Chemistry, Academic Medical Center, Amsterdam, 1100 DE, The Netherlands

PARENTS' PLACE

American College of Emergency Physicians

The American Academy of Pediatrics and American College of Emergency Physicians have developed a two-page Emergency Information Form for Children with Special Needs. The form is designed to be filled out by the child's doctor as a precaution before a crisis arises. When presented during an emergency it can give the emergency professionals a quick overview of the child's needs. The form is available online at:

<http://www.acep.org/2,468,0.html>

"I know for me, every time we go to the Emergency Room it is very frustrating because each time we have to explain to each new doctor what Barth syndrome is and what the symptoms are. This form will help to give the highlights of our child's disease. I also feel it is good to have the highlights of this disease written on paper so that we don't forget to mention any important details that we might otherwise forget to mention during this stressful time."

Submitted by Sheila Mann

A connection was made...

"The first article about Barth syndrome was written in 1981 by Dr. Barth and in that article he gave a description of a large family with a long history of children affected by this syndrome. That large family was my family. And so has my Barth boy Peter been guided by Dr. Barth since 1989. In 1999 Dr Barth told me that some parents of Barth boys in America were active on the Internet. I didn't have Internet myself, but there was a computer for public use in our village. So I used that computer to search on the Internet for Barth syndrome. The only thing I could find was a message of Moira Masterson on a page for children with special needs which read 'Moira would like to have contact with Barth parents and grandparents.' I left my home address on the same message board with a note that I didn't have Internet, but that I would love to write with her by snail mail. Within two weeks I received 4 letters: of Moira, Anna, Sue and Shelley, a grandparent of a Barth boy, and the three woman who started our original support group, respectively. I felt deep emotions when I read their stories and warm words for me and my family. I felt very happy to have contact with families in the same circumstances as ours. The understanding and the attention and respect in all these letters was great!

Then I read about the plans for a convention in Baltimore. I thought 'I would love to go there and meet everyone in person but it was impossible for me to take my husband and Peter with me because of language problems, so I asked Nel Beemsterboer for company.' Nel has had a Barth boy who passed away in 1985. She is the daughter of the eldest sister of my mother. So we went together to Baltimore and it was an unforgettable experience. We both felt a strong bond with other Barth families at first sight, even before we knew for sure whether they were in Baltimore for the Barth convention. We were sitting in the lobby of the hotel and when a new family came in, we were telling each other: 'That is a Barth family!' or 'No, I don't think that that family will join our convention' and never did we fail in our judgments!

I cannot find the right words for all feelings I went through to see so many Barth boys together and see the strong bond they felt between them and to feel the strong bond with all parents and grandparents. The meetings and information we received about Barth syndrome (education, occupational therapy and the recent scientific research) were of great importance to me.

I'm so glad that I was there at the first Barth convention of the world! I want to give thousands of

(cont'd on page 19)

Sites of Interest

Preparing for Back to School

The American Occupational Therapy Association, Inc. (AOTA) ("<http://www.aota.org/>") has teamed up with retailer L.L.Bean, Inc. ("<http://www.llbean.com/>") to bring an urgent safety message to parents and schools. Increasingly heavy school backpacks are putting the nation's students at risk and may be causing long-term damage to their growing bodies, according to AOTA. "Every year, we're seeing more children with stooped shoulders, sore necks and aching backs from carrying school backpacks, and we can't afford to put our kids at risk for a lifetime of chronic health problems," says AOTA Executive Director Joseph C. Isaacs, CAE. Please visit their websites wherein you will find tips for parents and kids on choosing, loading and wearing backpacks, including:

- Choose a pack that is appropriate to the child's size and age.
- Load heaviest items closest to the child's back (never allow a child to carry more than 15% of his or her body weight).
- Wear both shoulder straps for an evenly balanced load (wearing a pack slung over one shoulder can cause a person to lean to one side and curve the spine).
- Adjust the pack so that it fits snugly to the child's back (A pack that hangs loosely from the back can pull the child backwards and strain muscles between the shoulders). The bottom of the pack should rest in the curve of the lower back; it should never rest more than four inches below the child's waistline.

Dreamsurfer Network

The mission of the *Dreamsurfer Network* Internet support and education network is to provide teenagers with life threatening illnesses a community of friendship and understanding. The *Dreamsurfer Network* links teenagers and local hospital Web sites from throughout the world in an encouraging, confidential, counselor supervised setting. The benefits of this network is to provide teenagers who have life-threatening illnesses the opportunity to communicate with other teens who are going through similar circumstances. This relationship provides the following benefits: problem solving, peer support, understanding, friendship, education, and fun. You may find this site at:

<http://www.dreamsurfer.org/>

A connection was made...

thanks to all who made this a reality. After the convention the website www.barthsyndrome.org came on Internet and I logged in weekly to get the latest news. I'm always excited when I get an update from our President or mail from Vice-President Anna Dunn or another Barth family member. It is very important and helpful for me to know about recent scientific research, to read about the experiences and questions of other families and to write about my own experiences. For example, a question was posted on Barth Syndrome's message board by a mom of a Barth boy regarding chickenpox asking if other families experienced any adverse reactions to the vaccine, and many parents responded with their experiences. Also, recently I received a suggested "Care Plan" for Barth boys in school that I found very useful. A lot of Barth families are so creative in solving problems and I can learn a lot from it. I would like to finish this letter with the latest arrangement with Peter's teachers: he is allowed to take tests for school earlier in the day and earlier in the week, when needed, separate from his classmates under view of his tutor so that he will not be too tired to get high scores. He is also allowed to take a large test in 2 parts with a break in between. This is a very helpful arrangement for him.

Warm greetings to everyone I met in Baltimore!

A Letter from Rosemary Baffa

April 24th of 2000 forever changed our outlook on Barth syndrome. It was on this date we realized how little we knew about Barth syndrome in comparison to what we do not know and what we are still striving to learn. Prior to April 24th of 2000, our son Kevin experienced the "typical" Barth syndrome manifestations. He struggled with all of them at various times throughout his life with varying severities. He was a typical "Barth boy". His clinical presentation may not have differed much from your son's. Those issues we did not know and for the most part, still do not know provides us with the driving force to push the envelope of Barth syndrome knowledge to a new level.

We wanted to share Kevin's experiences with you because we feel it is important for all of our boys.

On the Monday morning after Easter our children were restless. We decided to have an Easter egg hunt after breakfast. Kevin was right in there with all of them as we hid, found and double-checked all the eggs. As I began kitchen clean-up I heard my oldest son, Matt. "Mom, Kevin is blue!" He found Kevin face down on the floor. I called our emergency medical number 911. Ted began CPR. My neighbor, a nurse, continued CPR until the paramedics arrived. After emergent care Kevin was transported to the local hospital where he was put on life support and later transported to DuPont Children's Hospital. We were informed that Kevin had suffered a cardiac arrest. Due to the uncertain amount of time Kevin was deprived of oxygen we were told the next 48 hours were critical. We clung to our faith and made contact with Shelley Bowen, Sue Wilkins and Anna Dunn. Even after the initial 48 hours of danger we were told Kevin's life was still in a precarious state.

Still reeling in shock we needed to know "How could this have happened?" This was not a part of Barth syndrome, as we understood it. Our cardiologist called Dr. Kelley, who in turn called Dr. Barth. We learned there were no episodes like this in the U.S., however there were two boys in Europe who experienced this. We also learned that these boys did not survive. Kevin slowly stabilized enough to have an internal cardiac defibrillator placed in his chest for his safety.

Kevin was released from the hospital but had a long summer of rehabilitation to get through. Physical, cognitive and occupational therapy, as well as neurological, gastrointestinal and cardiology check-ups were the order of the day. We got through this with continual physical improvement in all areas but his heart. We are so thankful to all of you for your support. We are also thankful for Kevin's wonderfully cooperative spirit.

In September all things were anew again. Kevin was doing well, even attending school for half-a-day when he started with diarrhea. The diarrhea weakened Kevin. He lost more weight, which he could not afford to lose. This continued to perplex us as well as his attending physicians. He tested positive for C-diff and was medicated several times. The cardi-

ologist found Kevin had been having many episodes of high and irregular heartbeats. One episode was just a breath away from full V-fib again. The ICD was working! However, again no explanations of "why?" Upon follow-up testing we learned the C-diff was gone however the symptoms remained. The diarrhea is still being treated as a "red-herring" in the bigger picture of Barth syndrome. We continue to go along with new knowledge, trial-and-error, and more than anything, hope!

Love and prayers from our house to yours,

GRANDPARENTS' CONNECTION

"I am Derek Sedefian's grandmother", wrote Joyce Lochner from Albany, NY. My daughter had finally purchased a computer and her first words typed were "Barth syndrome", and there, for the first time, she found a support group for Barth syndrome. Lynda gave me the site so that I could visit it myself. The first thing I saw was "sign Guest Book". I didn't know what to put down so I simply said "My name is Joyce and I am Derek Sedefian's grandmother". The very next day via e-mail I received "Hi Joyce, I am Travis' grandmother", which began my journey with the Barth "family". I went to the first convention in Baltimore and met Travis' grandmother, Moira!!!! I had finally found someone who had walked the same path as I had. There were other grandparents at the convention and it was wonderful to meet many of them. I look forward to meeting more of the grandparents in person at the 2002 convention. We have great hopes that more grandparents will want to become involved!!!! We are hoping that at the next convention we will be able to have a "special time" for us grandparents to be able to connect with each other!! It will be nice to see everyone again, and we are hoping that by 2002 our "grandparent group" will be larger.

Submitted by Joyce Lochner

Calling all News... We need to hear from you! This is your newsletter so please let us know what information you would like to see within this document. Please feel free to submit your articles to Lynda Sedefian, News Editor, at: lsedefian@barthsyndrome.org or by snail mail to 31 N. Grandview Terrace, Voorheesville, New York 12186.





The Heart of the Matter...

A special place for our young men and children to correspond with others around the world

An interview with 14 year-old Michael Bowen with Barth syndrome

Why is BSF important to you, as a boy with Barth syndrome?

"To begin with I have hope and believe that a cure will someday be found for the disease that I have. Without this group I would never have known any other boys with the same disorder. I have learned more about Barth syndrome from doctors and families who have either treated this disorder or have lived with it! When someone says 'I know how you feel' I know they mean well but they have no idea how I feel. When someone who has Barth syndrome says that 'I know', they REALLY do know how I feel."

Why is it important to know other boys with this disorder?

"It is important to me to meet other boys because it gives me an idea of what they are going through and I do not feel so lonely anymore. It's difficult to go to the cardiologist and know there are no other boys with the same disorder or not be able to connect with others just like me. Until mom started working with this group we thought I was the only boy in this country living with Barth syndrome. Now I know boys from all over the world and not only do I feel less lonely, but I feel I have met friends that will be there for a lifetime."

Why do you want to go to the BSF conference?

"Because I get to visit with my new friends, see Dr. Barth (who is AWESOME!) and the real reason is that it is FUN!"

What would you say to the Foundation?

"Keep working for us, there are a lot of kids depending on you!"

What would you say to doctors involved in the Foundation?

"Keep looking for a cure! Everyone is depending on you!"

Hey Kids....

Please write to us and share your concerns, stories, jokes, etc. You may submit your articles to Lynda Sedefian, News Editor at lsedefian@barthsyndrome.org or by snail mail to Lynda Sedefian, 31 N. Grandview Terrace, Voorheesville, New York 12186. We'd love to hear from you!

Michael Bowen has asked that other kids try to hook up with him on MSN to enjoy the opportunity of playing some fun games together. Please contact Michael at pickle_eater12@barthsyndrome.org for more information on how to go about connecting with him to have some fun!



Our mission is to guide the search for a cure, to educate and support physicians and to create a caring community for affected families.



THE BARTH SYNDROME FOUNDATION, INC.

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