Barth Syndrome Foundation
www.barthsyndrome.org

NEWSLETTER

VOLUME 2, ISSUE 1 APRIL 2002

World Class Scientific and Medical Advisory Board Established

Katherine R. McCurdy - ex officio
Vice President, Science and Medicine

The Barth Syndrome Foundation (BSF) achieved a very important milestone recently with the establishment of a top-notch Scientific and Medical Advisory Board (SMAB). This group will help educate us and the medical community about Barth syndrome, will share thoughts about improvements in treatments, and will help us further research that will lead ultimately to a cure. In short, the SMAB will be a critical element in our quest to "save boys' lives through education, advances in treatment and pursuit of a cure." This team has been carefully selected to represent all the fields most closely associated with Barth syndrome; its members span clinical specialties (such as cardiology, neurology and hematology) as well as areas of research expertise (such as biochemistry, molecular genetics and lipid metabolism). In addition, it is an international group that represents an array of world-class institutions. We are very pleased to have received such resounding support and participation from those whom the Board of the BSF invited to serve on this team, and we are proud to officially announce its initial membership:

Richard I. Kelley, M.D., Ph.D. – Chairman
Director, Division of Metabolism, Kennedy Krieger Institute
Associate Professor, Department of Pediatrics, Johns Hopkins University
Baltimore, MD

Dr. Kelley is an expert in metabolic diseases and has been involved in the treatment of more cases of Barth syndrome than any other individual in the US. He hosted the first International Scientific and Family Conference on Barth Syndrome in June 2000 at Johns Hopkins' Kennedy Krieger Institute. *(Continued on page 2)*

2nd International Scientific & Family Conference

Anna Dunn
Vice President and Family Liaison

It is with great joy and excitement that I write and personally invite you, on behalf of the Barth Syndrome Foundation, Inc., to attend our upcoming 2002 Barth Syndrome Family Conference which will be held at the Holiday Inn located in the Inner Harbor of Baltimore, Maryland on October 18th - 21st, 2002. In an effort to give our affected families the tools for becoming the most effective advocates for their children, this conference will provide the most up-to-date information on the forefront of research and resource information about Barth syndrome.

Our first-ever Barth Syndrome Family Conference which was held in June of 2000 was a huge success on many levels. Families bonded with other families and shared their little piece of the Barth syndrome puzzle. *(Continued on page 5)*

Inside this issue:

Published semi-annually by the Barth Syndrome Foundation, Inc, a 501(c)(3) non-profit organization dedicated to guiding the search for a cure, educating and supporting physicians and creating a caring community for all affected families.

All submissions and correspondence regarding the newsletter should be directed to:

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Back issues are also available.

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World Class Scientific and Medical Advisory Board Established

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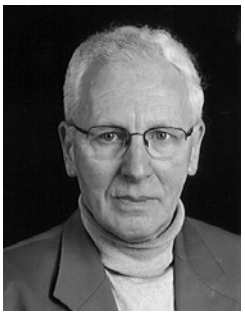

Peter G. Barth, M.D., Ph.D.
Pediatric Neurology
Emma Children's Hospital / AMC
Professor, Pediatric Neurology
University of Amsterdam
Amsterdam, The Netherlands

Dr. Barth is the pediatric neurologist who first described the condition that now bears his name, Barth syndrome. He has published extensively on the disorder and has treated a number of boys with Barth syndrome, many of them from the same extended family in the Netherlands.

Mary Ann Bonilla, M.D.
Division of Pediatric Hematology Oncology
St. Joseph's Children's Hospital
Paterson, NJ
Assistant Professor, Department of Pediatrics
Columbia University
Advisory Board, Severe Chronic Neutropenia International Registry

Dr. Bonilla was involved in conducting the initial clinical trials for G-CSF as a treatment for congenital neutropenia while at Memorial Sloan-Kettering Hospital. She is a pediatric hematologist oncologist who maintains an active interest in the treatment of white cell disorders, including Barth syndrome. As an Advisory Board member for the Severe Chronic Neutropenia International Registry, she has helped treat at least one Barth patient and has been involved with data concerning many more patients with Barth syndrome.

Gerald F. Cox, M.D., Ph.D.
Medical Director, Department of Clinical Research
Genzyme Corporation
Cambridge, MA
Assistant in Medicine, Division of Genetics
Children's Hospital
Boston, MA

As a clinician in genetics, Dr. Cox has been involved in the care of several Barth patients. His particular interests include the genetic basis of cardiomyopathy and treatment of inborn errors of metabolism. In addition, he oversees clinical trials for a well-known biotechnology corporation.

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World Class Scientific and Medical Advisory Board Established

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Salvatore DiMauro, M.D.
Lucy G. Moses Professor of Neurology
Columbia Univ. College of Physicians and Surgeons
Director, Columbia University Myopathy DNA Diagnostics Laboratory
New York, NY

Dr. DiMauro is a world-reknown authority on mitochondrial diseases. His laboratory conducts biochemical and DNA investigations of human metabolic myopathies.

Annette Feigenbaum, MB ChB, FRCPC
Division of Clinical and Metabolic Genetics
The Hospital for Sick Children
Assistant Professor, Department of Pediatrics
University of Toronto
Toronto, Ontario, Canada

Dr. Feigenbaum is a clinician who has helped treat a number of Barth patients in Canada. She also is a researcher whose primary interests include mitochondrial diseases.

Iris L. Gonzalez, Ph.D.
Molecular Diagnostics Laboratory
A. I. DuPont Hospital for Children
Wilmington, DE

Dr. Gonzalez works in the only molecular diagnostics laboratory that has been approved by the CLIA (the Department of Health and Human Services' Clinical Laboratory Improvement Amendments) in the US to conduct the genetic analysis necessary to confirm a diagnosis of Barth syndrome. She has written a layman's guide to genetics that has been extremely valuable to BSF families and others.

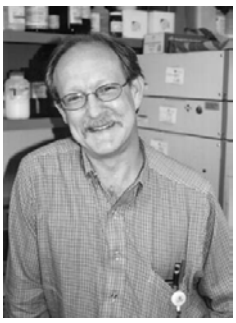
Grant M. Hatch, Ph.D.
Director of the Lipid Lipoprotein and Atherosclerosis Research Group
Professor, Dept. of Pharmacology and Therapeutics
Department of Biochemistry and Medical Genetics
University of Manitoba
Winnepeg, Manitoba, Canada

Dr. Hatch's research interests focus on metabolism and pharmacological modulation of phospholipids (including cardiolipin) in the mammalian heart and cells in culture. He has published numerous papers on these topics.

Michael Schlame, M.D.
Department of Anesthesiology
NYU School of Medicine
New York, NY

Dr. Schlame has had a long-term research interest in cardiolipin. Recently, this has led him to do some work on Barth syndrome specifically.

Jeffrey A. Towbin, M.D.
Associate Chief, Pediatric Cardiology
Professor, Department of Pediatrics and Molecular and Human Genetics
Baylor College of Medicine
Director, Phoebe Willingham Muzzy Pediatric Molecular Cardiology Laboratory
Texas Children's Hospital Foundation Chair of Pediatric Cardiac Research
Director, Heart Failure and Transplant Program, Texas Children's Hospital
Houston, TX



Dr. Towbin is a pediatric cardiologist whose major interests include cardiomyopathy, cardiovascular genetics and cardiac transplantation. In addition to his clinical practice, he also conducts research in related areas.

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World Class Scientific and Medical Advisory Board Established

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Ronald J. A. Wanders, Ph.D.
Laboratory of Genetic Metabolic Diseases
Academic Medical Center
Professor Dr., Department of Pediatrics
Emma Children's Hospital
and Department of Clinical Chemistry
University of Amsterdam
Amsterdam, The Netherlands

Professor Wanders heads the laboratory in which the very interesting recent work concerning the underlying biochemical causes of Barth syndrome was conducted. This laboratory works very closely with Dr. Peter Barth.

Katherine R. McCurdy – *ex officio*
Vice President, Science and Medicine
Barth Syndrome Foundation, Inc.
Larchmont, NY

Mrs. McCurdy and her husband are founding members of the Board of Directors of the Barth Syndrome Foundation, Inc. (BSF) where she serves as Vice President of Science and Medicine. As the mother of a son with Barth syndrome who was born in 1986, she is strongly committed to the goals of the BSF. She has an MBA from the Harvard Business School, with a concentration in management.

The purpose of this up to 16 member group is "to advise the Board of Directors of BSF on scientific and medical matters and to assist with the educational and research activities of the organization." To accomplish this, the duties of the SMAB are multi-faceted and include (but are not limited to) such things as:

- keeping BSF abreast of developments related to Barth syndrome,
- developing research strategies that lead most effectively toward treatments and a cure for Barth syndrome,
- establishing and spearheading our grant review process,
- writing and evaluating printed material for BSF,
- guiding our attempts to increase awareness of Barth syndrome within the medical community,
- helping us improve our Barth patient survey,
- responding to scientific and medical inquiries,
- acting as spokespeople and ambassadors for BSF,
- participating in our conferences.

"We all owe a real debt of gratitude to these fine professionals who have agreed to serve on the SMAB. We certainly appreciate their knowledge, guidance, support and participation."

Terms of SMAB membership are four years (with the Chairman being elected to the leadership position for two years), but individuals may be invited by the Board of Directors to serve for additional terms. SMAB members are not compensated for their time or their expertise, (though reasonable expenses will be reimbursed). We all owe a real debt of gratitude to these fine professionals who have agreed to serve on the SMAB. We certainly appreciate their knowledge, guidance, support and participation. There still is much that remains to be done to conquer Barth syndrome and to create ***"a world in which not one more child will suffer or perish from this disorder,"*** as the BSF mission states, but with many dedicated Barth families, contributors and SMAB members working together, we have a chance to make a huge difference.

Announcing for the first time….BSF Funds Available for Barth Research

The Barth Syndrome Foundation, Inc. is very pleased to be able to offer funds for research. We will award up to a total of $100,000 by year-end 2002 to support high quality work with a specific focus on clinical or scientific aspects of Barth syndrome. We expect that many of the projects submitted will be for the collection of preliminary data or the testing initial hypotheses. As a result, we expect that the $100,000 will be split between a number of grants that may range from $10,000 to $40,000.

We will have a two-stage grant review process in which initial Letters of Intent are to be submitted for review by the BSF Scientific and Medical Advisory Board. Based on this, the Board of Directors will then invite a number of investigators to submit full Grant Applications. The deadline for submission of Letters of Intent from interested investigators will be **June 30, 2002**. Please watch our website (www.barthsyndrome.org) for further grant guidelines and application process details; these will be posted by April 29, 2002.

2nd International Scientific & Family Conference

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On a scientific level, physicians and researchers interested in many facets of Barth syndrome collectively shared the most up-to-date information, which has stimulated an increased interest in ongoing research. Also, as a result of this conference, and most importantly, our "group" set out on its journey to become "The Barth Syndrome Foundation, Inc.", a 501(c)(3) not-for-profit organization.

The 2002 Barth Syndrome Family Conference format will be a bit different than the previous one. The family meetings will have a *panel format*, focusing on several specific areas of interest, concern, and relevance to those affected by Barth syndrome. The panel will consist of various specialists on Barth syndrome. These specialists will introduce their topics by giving a brief introduction to their specialty and then open up the sessions with questions from the families, followed by responses from the specialists. This format will offer our families a more intimate interaction with these specialists to promote a deeper understanding of the many complex issues of Barth syndrome. In bringing together the "family portion" of this conference with the "scientific portion", there will be a combined "group meeting" with all Barth families and physicians/researchers. This "group meeting" will educate the families in that it will offer the most up-to-date scientific information known about Barth syndrome. It will also allow our families an opportunity to address specific concerns to those physicians/researchers who will be in attendance. In addition to this "group meeting", there will also be specific blocks of time allotted for families to schedule an individual consult with various specialists of this disorder.

All Barth syndrome family members, grandparents, and other interested individuals are invited to attend. Siblings are definitely welcome and will enjoy the same activities as the Barth boys. This conference is for the whole family…so come and enjoy this experience of sharing and learning! You will be glad you came! The Holiday Inn, Inner Harbor is wheelchair accessible throughout the entire hotel. It also has an indoor heated swimming pool so don't forget to bring your bathing suits!! Social activities have been planned throughout the conference. The Starlight Foundation is planning a social night for the Barth Syndrome families and physicians/researchers. More details will be given in the upcoming months.

It is imperative that you reserve your rooms ASAP to ensure accommodations at the Holiday Inn, Inner Harbor. If you have an interest in attending this upcoming 2002 Barth Syndrome Family Conference you may contact Anna Dunn, Vice President and Family Liaison, at **adunn@barthsyndrome.org**. For your convenience, registration forms can be filled out directly from our website at **www.barthsyndrome.org**. Please fill out the registration form ASAP.

Important Steps to Take Prior to 2002 Conference::

1. Fill out 2002 Barth Syndrome Registration form and return it in the self-addressed envelope to Anna Dunn ASAP.
2. Reserve your vacation time for the upcoming 2002 Barth Syndrome Family Conference.
3. Call the Holiday Inn today and reserve your room at the $94.00 (plus tax) special rate. Rooms will be given to families on a first call, first serve basis…so **call today**!! **1-800-HOLIDAY** (USA and Canada only) or **1-410-685-3500** (from any location). Be sure to mention you are reserving rooms for the Barth Syndrome 2002 Family Conference!
4. Ask your son's physicians to reserve their schedule in advance if they are interested in attending the upcoming 2002 Barth Syndrome scientific portion of this conference which will be held in conjunction with the 2002 Barth Syndrome Family Conference. Dr. Richard Kelley is hosting the scientific portion of this conference, and interested physicians and scientists will be invited to attend those meetings. Dr. Kelley's meeting will take place at the same time as ours, and we will hold some sessions jointly so that the families, physicians, and researchers will all have the opportunity to meet together.

I can't wait to see you all there and share in our common grounds, obtain the most up-to-date information about Barth syndrome from various distinguished Barth syndrome physicians/researchers, and unite as "one family". Barth children will have the opportunity to connect with other Barth children, siblings will connect with other siblings, mothers and fathers will share their piece of the puzzle on Barth syndrome, and grandparents will connect with other grandparents and visualize the hope for a better and healthier tomorrow for our present Barth children and future generations.

We need you to join and share your piece of the Barth syndrome puzzle so that we can all move one step closer to potential treatments and a cure. If you have any further questions, do not hesitate to contact me at **adunn@barthsyndrome.org** and please bookmark our website at **www.barthsyndrome.org**, where you will find the latest information pertaining to this conference. See you there!!

First Calendar Year Accomplishments of The Barth Syndrome Foundation, Inc.

Shelley Bowen, President

The publication of this newsletter marks our first year as The Barth Syndrome Foundation, Inc. Because of this we wanted to show our constituency just how far we have come.

In 1996 my family, like so very many of you, thought we were alone in dealing with this disease. In 1998, we began to informally find other family members via the Internet. In 2000, we as parents sitting on the floor in a hotel conference room, voted to form a non-profit organization to better advocate for our collective concerns about our children. In the latter part of 2000, we incorporated as The Barth Syndrome Foundation. In early 2001, the Board of Directors held a retreat to organize our ideas into a comprehensive, well thought out strategic plan. Every detail was scrutinized. We knew we had to be realistic about what we can accomplish, but we knew that we would not waiver from our ultimate vision...***A world in which not one more boy would suffer or perish from Barth syndrome.***

Since that meeting, we have grown from a small group of concerned parents to a growing organization with an organized mission. We have our sights on a vision to see this life-threatening disorder as a treatable, and ultimately a curable disorder. Our vision is no different than it was when each of us came to know the words of Barth syndrome. But we are no longer alone and we are no longer in the realm of a small support group. We now are in the world of funding trying to raise money for research, in the world of strangers trying to teach them about this disease, and in the world of parents letting them know we are here to pave a road toward making a difference in all of our lives.

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Barth Syndrome Foundation Has a New Look!

Steve McCurdy, VP, Planning & Development

Thanks to the pro bono assistance of Barbara Sullivan and her company Sullivan & Co., a Madison Avenue marketing and consulting firm, we have a New Look! At the request of the Board, Barbara – whose clients include AOL Time Warner, Better Homes and Gardens, Fidelity Investments, Pfizer Foundation – agreed to create a new graphic identity for the Barth Syndrome Foundation, which would be uniquely ours. An organization's identity encompasses the design of its logo, its choice of type-face and color, the layout of all of its printed and other materials – in short, the way in which it chooses to represent itself.

When we created the foundation and drafted our vision, our mission and our strategic plan, the Barth Syndrome Foundation, Inc. became our legal name, as announced in the last newsletter. None of that is being changed. However, while our previous logo was a wonderful evocation of our focus on family, it was created out of components that are all in the public domain… which means anyone could copy them. We decided we needed a distinctive design for our logo and a look for all of our correspondence that would immediately identify us as the Barth Syndrome Foundation and remind all of our constituencies of who we are and what we stand for. The new Barth Syndrome Foundation graphic identity program was conceived as a key part of our effort to raise the visibility of this disease and increase awareness among all of those we serve and hope to reach: the medical community; families who are affected by the disorder; contributors; and volunteers.

Peer Review: As a first step, Sullivan & Co. conducted a "market analysis" to understand and evaluate graphic identity systems for similar non-profit organizations. Based on this research, several conclusions and guidelines emerged:

- Simple worked better than complicated

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Barth Syndrome Foundation Has A New Look!

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- The number of colors (and therefore the cost of production) didn't seem to influence overall effectiveness; simple one-color materials looked as good as four-color designs. To reinforce this idea, we noticed that some of the largest, best funded, and most successful organizations had the simplest identities.
- The name of the organization was more important than the symbols and designs (which were often difficult to decipher in small formats anyway).

Design Criteria: After extensive discussions with the BSF Board, Barbara decided that the identity had to fulfill several objectives:

- It had to appeal to all four constituencies.
- It had to convey the professionalism and seriousness of our foundation, but it also had to clearly remind everyone that our primary focus is still the children – the boys who live with Barth Syndrome every day.
- It had to symbolize the condition as well as signify hope and our thoughts for the future.
- It had to boldly communicate the name of our organization for maximum visibility when included among those of many other groups and foundations.
- It had to work in one-color and black-and-white formats to minimize production expense and to simplify

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2001 Development Report

Steve McCurdy, VP
Planning & Development

We started the year off with good news from the IRS (yes, it does happen occasionally!) – our application to become a tax-free, not-for-profit organization (a "501 (c) (3)" entity, for the tax savvy among you) had been granted! This meant that any donations to the Barth Syndrome Foundation, Inc. would be tax-free to the donor… and that BSF would not have to pay income taxes as long as we continue to meet the standards set by the IRS.

In our application, we had to describe our foundation's purpose, and the programs we expected to run. We had to submit a copy of our official incorporation documents, by-laws, a legal opinion and a list of our board members. We also had to project our financial statements for three years indicating how we would raise money and how we would spend it. The Board of Directors set a goal of raising $785,000 over the next five years.

The IRS requires that BSF file a return each year detailing how we raised money and how we used it to make sure that we continue to operate as we said we would. Since we are a public charity, they take particular interest in how, and from whom we have raised money, and they want to see at least 33% coming from a broad base of donors. Excluded from this list is anyone who donated in excess of 2% of total contributions.

We did quite well in our first full year as a charity. We raised a total of $145,327 from over 300 contributors. As you know from our last newsletter, we received a single grant of $100,000 from a private foundation with the help of the Baffa family, and several donations in excess of the 2% mark of $3,000. So excluding those larger donations, we will report to the IRS that we raised 21.3% of our total donations from smaller contributions. Over the next two years we must raise that figure to 33% on average, or request a legal exception to this requirement.

How did we do it? Well, Lynda Sedefian raised over $15,000 from her Barth Syndrome Walkathon in her hometown of Voorheesville, NY. The Baffa family raised almost $2,000 at a holiday party, and Cherie Schrader raised $1,500 by asking sponsors to back her as she ran in the Chicago Marathon. We also raised over $25,000 from the BSF Board of Directors and Barth families and friends, including a $10,000 founding donation from Paula Varner, Sue Wilkins' mother.

We are well on our way to another successful fund and aware-

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First Calendar Year Accomplishments ...

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During our first year of existence, we understood the importance of strategic planning if we intended for this group to be a driving force for Barth syndrome awareness, research and family support. Using the five primary program goals we developed as guidelines, we have stayed the course to develop into a stronger organization for our constituency. Below is a list of our accomplishments toward our five stated goals during our first year.

Program Development

1. ***To insure that all appropriate physicians are aware of Barth Syndrome, have ready access to the latest information to insure an accurate diagnosis and can easily make use of the medical resources they need to deliver successful treatment.***

a. Developed BSF website with www.barthsyndrome.org domain name to serve as central location for information about Barth syndrome.

b. Posted most comprehensive database of Barth articles on BSF Website with a variety of formats for retrieval. Developed Barth informational brochure for ready access to symptoms and history of Barth syndrome.

c. Worked with Dr. Richard Kelley to create a document identifying a step-by-step procedure to diagnose Barth syndrome. This is now posted on the BSF website.

d. Identified physicians with expertise in Barth syndrome and posted their names and contact information on the BSF website for expedient accessibility for remote consultative needs.

e. Worked directly with Iris Gonzalez of the A. I. DuPont Laboratory to contact physicians and, ultimately, affected families with a positive diagnosis of Barth syndrome.

f. Spearheaded effort to have Barth syndrome included in the latest edition of the NORD Directory.

g. Automated Barth survey so that results can be tabulated more easily and data can be used more effectively.

h. Held advisory meeting with key members of Columbia Univ. College of Physicians & Surgeons representing multiple pediatric specialties to define and develop an improved physicians awareness program.

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

a. Established the BSF Scientific and Medical Advisory Board with an initial membership of eleven highly regarded international members representing various clinical and scientific specialties that are particularly relevant to Barth syndrome and one ex officio member from the BSF Board of Directors.

b. Facilitated research conducted by Dr. Michael Schlame on developing a new screening test for Barth syndrome.

c. Collaborated with Michele Mazzocco for research on Cognitive Development of children with Barth syndrome leading to preliminary findings of cognitive delays in children with Barth syndrome.

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

a. Developed brochure for parents focusing on effective ways of caring for a child with Barth syndrome

b. Developed a listserv community to serve as a forum for families to exchange vital information with one another for immediate access to informed opinions from fellow Barth parents and physicians.

c. Developed a private website for families to freely exchange photographs and stories without concern of predators.

d. Created a private chat room where families and affected boys can go for real time chats online free of charge to socialize.

e. Published first ever Barth Syndrome Foundation Newsletter with informative articles with our entire community contributing to the credibility of the document as a sound source of quality information.

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. Applied for and received 501(c)(3) tax-free status from IRS.

b. Registered in six states (FL, NY, NJ, CT, MA, IL) to permit fund raising and other activities.

c. Raised in excess of $145,000.00 from over 300 contributors ranging from major national foundations to families, friends and interested individuals from several countries.

d. Helped sponsor several fund raising events including community walkathons and marathon entrants and

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First Calendar Year Accomplishments …

(Continued from page 8)

sent two letter solicitations to Barth families and friends.

e. Accepted by Helping.org through which we can accept contributions on our website from contributors using all major credit cards.

f. Also registered with GuideStar, a national registry of charitable organizations seeking contributions and volunteers and affiliated with Helping.org.

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

a. Increased the Board from five to six members, adding Lynda Sedefian as Corporate Secretary.

b. Created a comprehensive living document clearly identifying our goals and objectives for the various constituencies of our community, and outlining programs to be implemented in the coming year.

c. Hosted two Board retreats to educate board members and develop programs and goals.

d. Developed official Barth logo and graphic identity and adopted a new tag-line.

e. Began to plan the Second International Barth Syndrome Scientific and Family Conference, to be held in Baltimore in October of 2002.

2001 Development Report

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ness raising year in 2002 as well. So far, in the first two months of 2002 we have raised over $87,000 from Board letter solicitations including two donations of $20,000 and $25,000 each.

Lynda, the Baffas, and Cherie have all shown us that individually we can make an important contribution to the Barth Syndrome Foundation by raising awareness and money in a variety of ways. They did it with walk-a-thons, marathons and holiday parties. You could help by turning something you love to do into a fund-raiser for the BSF. Love to play golf, go bass fishing or play bocce? Have a tournament! Do you walk, ride a bike or run for exercise? Gather your friends and sponsors and race for BSF! Is partying more your style? How about a house party? Or send a personal mailing out to all of your friends and family – we can help you compose the letter. There are a million ways to get involved and every time you do, you help increase awareness of Barth syndrome – one of our critical objectives, as well as raise additional funding for research, workshops, education and family support.

We can raise money for BSF in 18 states, with three more in process. A list of states where we can solicit accompanies this report. If you need help, let us know. We can help you write letters, organize a walk-a-thon, guide you on getting help within your community and much more. We are always happy to have volunteers! Please contact me at SMcCurdy@barthsyndrome.org.

Listed herein are the states in which Barth Syndrome Foundation Inc. can do fund raising: Colorado, Connecticut, Delaware, Florida, Hawaii, Idaho, Indiana, Illinois, Iowa, Massachusetts, Montana, New Hampshire, New Jersey, New York, Nebraska, Nevada, South Dakota, Wyoming, and Texas.

States in which Barth Syndrome Foundation Inc.'s registration is pending: California, Maryland, and Tennessee.

Chicago Marathon

I am Lattigo Cook's Aunt Cherie'. Lattigo was born on May 1, 2000 with Barth syndrome. Just as everyone else, I was very confused and frustrated by this illness. I felt like my hands were tied because I am a person that does everything I can to help my loved ones. I wondered how I would be able to help with very little knowledge, so my goal became to obtain knowledge and I was introduced to the foundation. Knowing Lattigo was having a difficult time with his muscles, I thought that I would use my muscles and physical activity to help. I decided that I would run the Chicago Marathon on October 7th for Lattigo and create awareness about Barth syndrome. Armed with passion and love, I hit my training schedule, and approached many businesses for sponsorship. Family, friends and strangers became a part of this fundraiser and were all willing to help. Without everyone's support it would not have been so successful.

BSF Has A New Look!

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reproduction in offline and online applications. From this basis, Barbara and her team developed a number of different options and reviewed them all with the Board. Together, we compared them to the criteria we had established and discussed their strengths and weaknesses. Eventually, we settled on the blue, spiral hand-drawn design you see here on the Newsletter masthead, and on all our new materials. And we agreed on the child-like lettering, which spells out Barth Syndrome Foundation, Inc. and reminds us of what we are all about.

The new Barth Syndrome Foundation identity gives a facelift to the organization and represents a focus on the future. The spiral design element reinforces the all-encompassing nature of the condition with a heart at the core – a symbol both of some of the muscles that are afflicted by this disease - as well as the hope and love that we give to all children who suffer from it. Bright blue was chosen as a color both for its boldness and strength, as well as to symbolize boyhood. After much discussion and editing, we also settled on our tagline that would become a brief, memorable version of our mission to be displayed prominently and frequently. Each word was selected carefully to convey the nature and importance of our cause and the hope embodied by our foundation.

Finally, the Barth Syndrome Foundation name is a key element of the graphic identity to give the organization maximum recognition. The logo and lettering and their use as design elements on our newsletter, brochures, letterheads, website and all the rest of our communications materials clearly indicate a professional approach worthy of respect and recognition. But the child-like style of both is an unmistakable reminder to everyone of our vision of ***"...a world in which not one more boy should suffer or perish from Barth syndrome."***

Those of us on the Board who oversaw this effort learned a lot about how to convey a complex message with a set of simple designs at minimum cost. It was wonderful to have Barbara Sullivan and her talented and professional team to guide us, and we very much appreciate their efforts to help give us

Tax-Deductible Donations to Support BSF may be made via :

Barth Syndrome Foundation, Inc. P.O. Box 23173 Lincoln, NE 68542-3173	www.barthsyndrome.org (click on Helping.org button)

2001 Financial Report

Sue Wilkins, Treasurer

Accompanying this report are a Statement of Revenue and Expenditures and a Balance Sheet for the Barth Syndrome Foundation, Inc. for the year ending December 31, 2001. Following completion of our audit in March of 2002, we will file our returns with the IRS and the states in which we are registered. You will also be able to find our audited financial statements on our website at www.barthsyndrome.org.

We successfully raised over $145,000 in donations including a single purpose grant of $100,000 dedicated to research. Details of our fund raising efforts can be found in Steve McCurdy's Development Report. We also earned $1,771 in interest reflecting the conservative investments on our balance sheet. We were careful with our expenses as well, spending just under $7,000 in our start-up year. We spent only $484 on fund raising, under $5,000 on administration and $1,575 on programs, principally awareness. We ended the year with $140,849 in the bank, $100,000 of which is restricted to research only. These are excellent figures reflecting an all-volunteer organization and a first year focus on creating the foundation to allow us to grow and make a significant impact on our objectives in coming years.

While we are now in the process of developing our plans and detailed budgets for 2002, it is safe to say that expenditures on programs will increase as we sponsor our 2nd International Barth Syndrome Family and Scientific Conference in October, publish two newsletters, expand our awareness programs and begin the process of soliciting, evaluating and awarding research grants with the help of our newly formed Scientific and Medical Advisory Committee headed by Dr. Kelley. We have a well structured planning and budgeting process in place, we maintain close control over expenses and we will continue to invest the Foundation's assets with an eye to asset preservation and liquidity. We have built a cushion of investments, a portion of which we will maintain permanently as a source of liquidity. We will continue to be prudent in the management of our resources in 2002.

To support these programs we must continue to focus on and expand our fund raising efforts. We will repeat the fund raising activities that worked in 2001, but will need to enlist more people willing to sponsor more events and increase awareness in 2002.

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2001 Financial Report

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The Barth Syndrome Foundation, Inc. has created real momentum in 2001 – creating the organization, beginning the awareness, family support and research programs and creating a successful fund raising process. We are looking forward to making real progress against our long term objectives in 2002.

Barth Syndrome Foundation, Inc.
Balance Sheet as of December 31, 2001

Assets	2001	2000
Current Assets		
Unrestricted Funds	$ 40,849	$ 697
Restricted Funds	$100,000	$ -
Total Current Assets	$140,849	$ 697
Total Assets	**$140,849**	**$ 697**
Liabilities & Fund Balance		
Fund Balance	$140,849	$ 697
Total Liabilities and Fund Balance	**$140,849**	**$ 697**

Barth Syndrome Foundation, Inc.
Statement of Revenue and Expenditures
For the Year Ended December 31, 2001

	2001	2000
Revenue		
Grant Income	$ 99,992	$ -
Interest and Other Income	$ 1,806	$ 22
Donations	$ 45,335	$ 3,209
Total Revenue	**$ 147,133**	**$ 3,231**
Expenses		
Administration	$ 4,922	$ 737
Fund Raising	$ 484	$ 1,073
Programs	$ 1,575	$ 724
Total Expenses	**$ 6,981**	**$ 2,534**
Excess or (Deficiency) of Revenues over Expenditures	**$140,152**	**$ 697**

A Child's Educational Needs

Joan C. Stoner, Ed.D.

What should I be looking for with respect to my child's educational needs? First, and foremost, medical/health issues are primary. Education is secondary. Depending on your child's medical history, you must determine how far behind he may be than other children in comparison to the amount of time he has been available for learning and absorbing knowledge and information that children without his health issues.

If you have kept a written history of the number and length of times he has spent in the hospital from birth until his current age, it is reasonable to count those days, add 10% for the number of days he just wasn't up to par in, and subtract that

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Empowering Support Groups

Sharon Terry, President, Genetic Alliance

Welcome to The Barth Syndrome Foundation as a member of the Genetic Alliance! We are a coalition of 300 such groups –from large cancer groups to small, rare disease groups – working together to help each other to better serve our members. Our groups enlist the voices and engage the hearts of individuals and families who yearn for solutions, relief, services, treatments, comfort.

The Genetic Alliance was founded to help support groups – because they are where no one else would be. There are no other entities in our society that could or would rise to the task of giving voice and taking action on behalf of the millions of people living with genetic disorders. The power of lay advocacy groups lies in the simplicity and purity of their mission, their passion for their endeavors and clear vision of the task before them. Their power is forever practical and focused. It can move mountains, beginning in the mind of an anguished parent and growing to become a strong, effective organization.

The Barth Syndrome Foundation embodies the essence of the Genetic Alliance – empowering individuals and families to take needed action and make their composite voices heard. Most of all, and most importantly, the Barth Syndrome Foundation offers hope. This hope is a solid hope, forged from the stark reality of collective experience and the power and potential of people working together. The Genetic Alliance sees its mission as walking hand in hand with The Barth Syndrome Foundation and other groups, offering resources and assistance when needed, creating synergies between groups, and respecting the uniqueness of each group's challenge and solution.

Though the Barth Syndrome Foundation is concerned about a very specific genetic disorder, they join the Genetic Alliance in being a part of a massive consortium of people striving to improve the lives of individuals impacted by genetics. Under the umbrella of the Genetic Alliance, all these groups are empowered to face the smallest challenges and emboldened to overcome even more global and seemingly impossible obstacles. The Genetic Alliance is enriched by the membership of the Barth Syndrome Foundation, by the passion and hard work of Shelly Bowen for people affected by Barth Syndrome. We are here to help the Barth Syndrome Foundation in its mission!

THOUGHTS ON THE FUNCTION OF THE BARTH GENE

Richard I. Kelley, MD, PhD,
Johns Hopkins Medical Institutions

The initial concept of Barth syndrome as a disorder of mitochondrial metabolism was well-supported by laboratory data as well as by evidence of structural abnormalities of mitochondria in the muscle biopsies of Barth children. Moreover, some of the clinical problems of Barth syndrome, such as cardiomyopathy and muscle weakness, also occur in a variety of genetically well-defined disorders of mitochondrial energy metabolism. However, as we grew to know better both Barth syndrome and the full spectrum of mitochondrial diseases in the 1980s and 1990s, it became clear that Barth syndrome stood in contrast to all other mitochondrial syndromes because of several distinctive clinical problems, especially cyclic neutropenia and a severe growth retardation that largely resolves after puberty. In addition, quite a few children with Barth syndrome have had no clinically identifiable abnormalities of mitochondrial function or structure, such as lactic acidosis or abnormal-appearing mitochondria in heart or skeletal muscle biopsies. Also important was that studies of living muscle of two Barth syndrome boys in Philadelphia, despite especially severe growth retardation and cardiomyopathy, showed normal metabolism of ATP, the essential currency of energy that is the most important product of mitochondria. Thus, whereas most early research on Barth syndrome focused on understanding the mitochondrial energy metabolism of Barth children, thoughts were growing that Barth syndrome may be a more global metabolic disorder with major effects outside the sphere of mitochondrial function. However, where the primary genetic lesion was and how it might have extramitochondrial effects remained a mystery for many years.

"However, as we grew to know better both Barth syndrome and the full spectrum of mitochondrial diseases in the 1980s and 1990s, it became clear that Barth syndrome stood in contrast to all other mitochondrial syndromes because of several distinctive clinical problems, especially cyclic neutropenia and a severe growth retardation that largely resolves after puberty."

After much tedious but also insightful genetic study, the Barth gene and the proteins it produces (somewhat unfortunately named "tafazzins," after an Italian comedic character) finally were discovered and reported by Dr. Bione and her colleagues in 1996. However, much of the mystery of how that genetic lesion causes Barth syndrome remains to be discovered by the new Barth research that was begun by the late Dr. Peter Vreken and is now expanding to engage many other laboratories. Dr. Vreken's elegant experiments showed conclusively that the deficient Barth protein helps shape the final form of the mitochondrial-specific lipid, tetralinoleyl cardiolipin. In light of Dr. Vreken's recent findings, the original discovery of Dr. Bione and her colleagues that many different Barth proteins are made from the same gene now suggests an explanation for the many non-mitochondrial characteristics of Barth syndrome. In essence, the Barth protein is an enzyme, a "transacylase," that helps exchange one fatty acid for another in the synthesis of tetralinoleyl cardiolipin, a complex mitochondrial "phospholipid" made from glycerol, phosphorus, and four "linoleic" fatty acid molecules. However if cardiolipin is exactly the same in all mitochondria, why are there probably at least ten different Barth enzymes with different proportions in various tissues? Possibly, mitochondria in different tissues need slightly different Barth enzymes for cardiolipin biosynthesis, although a priori there is no reason to think that. Perhaps the answer is that other Barth proteins are involved in the synthesis of phospholipids that have roles outside mitochondria, such as in the cell membranes of neutrophils or the cartilage cells that determine growth, and that the Barth protein inserts linoleic acid into some of the hundreds of other lipid molecules that need linoleic acid.

The Barth gene is not unique in its ability to spawn multiple proteins. Indeed, thousands of individual genes encode not just one protein but multiple proteins through processes that, in effect, chop up and piece the gene back together in different ways. This process, called "alternative RNA splicing," is probably the main reason why humans, who are a hundred times more complex than a single-celled yeast, have only about five or six times as many genes as that microscopic organism's 6000. Not every human gene, but perhaps most, can be expressed as different proteins to serve related but different functions. Such gene plasticity may, in effect, double or triple the different number of protein products that a person's 30 to 35,000 genes can make. Moreover, even single proteins once

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How Barth Syndrome Research May Improve the Understanding of Other Human Diseases

Troy Phipps, Doctoral Student
Univ. of Southern California
Institute for Genetic Medicine

This article is intended to address questions on how Barth syndrome research may improve the understanding of other human diseases and basic science.

Several physicians and scientists were asked their opinions in the below questions, and we are fortunate that they agreed to share their fascinating visions and thoughts on this disease. It is not every day that we are allowed to peer into their minds to observe the scientific process at work, and to hear where future research directions could lead. From their collected responses it is clear that they consider Barth syndrome research an interesting and relevant topic not only for understanding more about our physiology and cell biology, but also clues as to how to diagnose new lipid remodeling disorders. I wish to thank Drs. Peter Barth, Richard Kelley, Michael Schlame, the Barth Syndrome Foundation and Kate McCurdy for their time and input.

What broader interests can come out of understanding the Barth Proteins and lipid remodeling in humans?

"Research on BTHS may help us to understand other forms of cardiomyopathy. It may also give new insight into the molecular physiology of the heart. Furthermore, the investigation of taffazzins may reveal information on the formation and remodeling of phospholipids. This is a fundamental problem that has essentially remained unsolved despite intense research efforts by many laboratories."

Dr. Michael Schlame

How might research results on Barth syndrome shed new light on other human genetic diseases? What specific example(s) come to mind?

"The control of cholesterol levels in the cell is a vital process. One way in which this is done is by regulation of its transport over cell membranes. This process is an important focus of research by many laboratories studying disorders as diverse as atherosclerosis, NiemannPick C disease (a neurodegenerative disorder), and familial hypercholesterolemias. In Barth syndrome, plasma cholesterol is low for still unknown reasons. One reason may be decreased synthesis, but this has not yet been confirmed. Recently it was discovered that in a rare disorder of cholesterol efflux (outward transport from the cell), named Tangier disease, cardiolipin is increased in the cell. Could it be a coincidence that in Barth syndrome plasma cholesterol is decreased, while there is a deficiency of cardiolipin? This raises the question whether a relationship exists between cardiolipin and cholesterol (transport). Study of cholesterol transport in Barth syndrome may become an important tool for our knowledge about cholesterol in general."

Dr. Peter Barth

"Research on BTHS may help us to understand other forms of cardiomyopathy. It may also give new insight into the molecular physiology of the heart."

"One of the most interesting questions is how a remodeling defect of cardiolipin can cause heart failure. Eventually the consequences of tafazzin mutations will have to be addressed on the cellular level and then integrated with cardiac physiology. In a similar fashion, BTHS research may help us better understand neutrophil maturation. Perhaps, neutrophils need very active mitochondria at some point in their maturation process."

Dr. Michael Schlame

How might understanding cardiolipin metabolism help doctors/scientists understand human biochemistry better?

"By studying cardiolipin metabolism we will understand more about the structure of mitochondria, an organelle which bears on nearly all areas of biological science."

Dr. Peter Barth

"Many of the most important clinical problems of Barth syndrome are not those of abnormal mitochondrial energy metabolism but, rather, those of abnormal growth---short stature during childhood, muscle hypoplasia, and inadequate production of neutrophils. The discovery that one or more Barth proteins is essential for the synthesis of a complex phospholipid, cardiolipin, has immediately focused attention on other possible disorders of phospholipid metabolism as causes of similar growth problems in other genetic syndromes. Like cholesterol, phospholipids are essential components of all cell membranes. It would therefore be expected that a deficiency of

(Continued on page 14)

Thoughts on the Function of the Barth Gene

(Continued from page 12) ...synthesized can be modified in a variety of ways in different tissues or even different parts of the same cell and thereby acquire further specialization of function, an aspect of Barth protein metabolism that has not even begun to be studied. Thus, part of the Barth research we eagerly await will focus on discovering all of the other, non-cardiolipin functions in lipid metabolism that we suspect this cadre of Barth proteins has. In this way, we hope to find treatments that improve not only energy metabolism in those Barth boys with mitochondrial problems, but also the more troublesome day-to-day problems of neutrophil production, skeletal growth, cardiomyopathy, and muscle development that affect nearly all boys with Barth syndrome.

"Thus, part of the Barth research we eagerly await will focus on discovering all of the other, non-cardiolipin functions in lipid metabolism that we suspect this cadre of Barth proteins has."

How Barth Syndrome Research May Improve the Understanding of Other Human Diseases

(Continued from page 13)
phospholipids as essential cellular building blocks would lead to deficiencies of growth. Thus, to find other defects of phospholipid biosynthesis among genetic disorders whose causes are unknown, scientists now know to examine first those conditions with Barth-like growth problems. The first of these disorders has already been identified in three children having a severe brain disease quite unlike Barth syndrome, but also having dilated cardiomyopathy and extreme short stature. Their biochemical studies suggest a defect not in the primary synthesis of phospholipids but in their recycling. Thus, for many genetic disorders with abnormal growth, and especially those that also have at least one of the cardinal features of Barth syndrome--dilated cardiomyopathy, neutropenia, muscle hypoplasia---we now have a rationale for identifying at least some of their genetic lesions and thereby developing new and more specific therapies for those children." ***Dr. Richard Kelley***

"One of the most interesting questions is how a remodeling defect of cardiolipin can cause heart failure. Eventually the consequences of tafazzin mutations will have to be addressed on the cellular level and then integrated with cardiac physiology. "

On behalf of the Barth Syndrome Foundation, we would like to extend our gratitude to Troy Phipps, as well as those physicians who participated in this interview, for their enthusiastic interest in Barth syndrome. Through all of our efforts, we continue to pave the way to a brighter future for those affected by Barth syndrome.

Genetics of Barth Syndrome

Iris L. Gonzalez, PhD, Molecular Diagnostics Laboratory
A. I. DuPont Hospital for Children

Barth syndrome is a condition with X-linked recessive pattern of inheritance". What does that mean? How was this pattern of inheritance determined? For a geneticist, the clue to pattern of inheritance was that only males were being affected and that, in large multigenerational families, the condition was transmitted through some females who had both affected sons and unaffected sons, while other females did not seem to transmit at all.

The sex of a person is determined by the sex chromosomes, X and Y. A woman has 22 pairs of "regular" chromosomes plus 2X chromosomes, and a man has the 22 pairs and 1X and 1Y. A woman is said to be 46,XX and a man is 46,XY. Now, in making eggs and sperm, a person only transmits one of each kind of chromosome: an egg has 22 + X (one or the other X), and a sperm has 22 + X or 22+ Y (the first would give rise to a girl, the second to a boy).

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GENETICS OF BARTH SYNDROME

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The gene that is responsible for Barth syndrome is called TAZ1 and is on the X chromosome. A woman has 2 copies of the X chromosome and therefore she has 2 copies of the TAZ1 gene; if one of her TAZ1 genes is mutated (and does not work) she will still be healthy because the one remaining gene makes enough product. The product made by this gene is an enzyme involved in processing lipids (fats) that are needed for healthy membranes in the mitochondria (the powerhouses of the cell). The pattern of inheritance is called X-linked (meaning that the gene is on the X) recessive (meaning that the mutated gene "recedes" or hides behind a normal one that makes enough enzyme). However, if a male has a mutated TAZ1 gene on his single X chromosome, he has no good backup copy like the female does, and he will suffer from Barth syndrome. This is why the condition is seen in males, while being transmitted by healthy females.

Transmission probability: a female that has one mutated TAZ1 gene and 1 normal TAZ1 gene can transmit either one in an egg, which means she has a 50% probability of passing the mutated gene each time she has a child. If the child is a female, the girl may be a "carrier" like her mother or may receive the normal copy and be a non-carrier, and if the child is a male he has a 50 % probability of receiving the normal copy and being healthy or receiving the mutated copy and being affected.

NEUTROPENIA IN BARTH SYNDROME: CLINICAL COURSE AND TREATMENT OF NEUTROPENIA

C Zeidler, PG Barth[2], MA Bonilla, AA Bolyard, L Boxer, T Cottle, DC Dale, J Donadieu, C Fier, M Freedman, G Kannourakis, S Kinsey, B Liang, B Schwinzer, K Welte, B Cham, for the Severe Chronic Neutropenia International Registry (SCNIR), Seattle WA, USA; Hannover, Germany, and [2] Department of Pediatrics, University of Amsterdam.

Barth syndrome (BTHS) is a rare X-linked recessive disorder characterized by cardiac and skeletal myopathy, variable neutropenia, increased 3-methylglutaconic aciduria, and short stature. The gene responsible for Barth syndrome, G 4.5, is located on Xq28 and has recently been cloned. Female carriers have a normal phenotype. BTHS is a severe disorder with a fatal outcome often during childhood due to cardiac failure. Neutropenia in BTHS syndrome has been described to be considerably variable with some patients being neutropenic during infectious episodes only. Most patients suffer from persistent oral infections, however, life-threatening infections in BTHS patients have also been reported. Based on accumulated data from the SCNIR we describe the course of neutropenia, history of infections and treatment for neutropenia in 7 BTHS patients reported to the SCNIR and one additional patient (AP) reported by Barth et al, 1983 (J Neurol Sci 1983; 62: 327-355). Patients' age ranges from 1 to 12 years. All patients reported to the SCNIR are alive and receive G-CSF treatment for severe chronic neutropenia. Onset of neutropenia was documented during the first months of life in 5 of the 7 SCNIR patients and the AP, and at the age of 2 or 3 years respectively in 2 patients. G-CSF was started between the third month and fifth year of age in the 7 SCNIR patients. All 7 patients receive G-CSF maintenance treatment with a dose range between 3.8 mcg/kg/day and 8.3 mcg/kg twice a week. None of the patients had a history of severe infections despite a documented absolute neutrophil count (ANC) below 500/µl prior to G-CSF treatment. While on treatment all 7 patients had a significant rise in their ANC. No severe infections occurred on maintenance treatment. The follow-up time ranges from 1 to 12 years (mean 5.7 years). In contrast, the patient originally described in 1983 (AP) showed only intermittent neutropenia and did not receive G-CSF treatment. This patient died from cardiac arrest during a febrile infection of the upper respiratory tract at the age of 15 months. Our analysis shows that patients suffering from severe chronic neutropenia due to BTHS respond clinically to G-CSF treatment. However, cardiomyopathy leading to cardiac failure is the limiting problem in the majority of these patients.

THROUGH THE EYES OF A GRANDPARENT…

Hi, Grandparents of all our boys everywhere. My name is Moira Masterson, Grandparent Liaison of the Barth Syndrome Foundation and the proud grandmother of Travis who will be seven in April. He is in grade one, has big blue eyes, a wonderful personality and loves his Granny's chicken soup. Travis has Barth syndrome.

It has been my privilege over the past three and a half years to have met many grandparents by way of the internet, "snail mail", phone, and in Baltimore, during June 2000, at the first ever Barth syndrome conference, whose lives have been touched by Barth syndrome. I have made many wonderful friends, shared many memories, feelings, tears, laughter and things I had never shared before with another living being.

For more than forty two years I thought I was the only one who had ever lost a little boy for no understandable reason. I was wrong. I now know that there were many others who suffered the same pain that I did, and who asked themselves the same question "Why?" We are now all grandparents and for those whose lives were affected by the Barth gene the answers are beginning to become clearer.

Being a grandmother is totally different from being a mother. When my children were born I experienced a love for them for which I never knew I was capable. I am certain you all felt the same way....but…

The love and devotion I gave to my child
I thought I could give to no other
But life had a pleasant surprise in store
One day I became a Grandmother
And how we love our grandchildren!!!!

When my grandson was diagnosed with Barth syndrome and I realized the implications, I felt as if my whole world had again fallen apart. The pain that I felt for my daughter and son-in-law could not be described, nor could it be compared to the pain I felt on losing my son. ***That*** was my pain. ***This*** was my children's pain. As parents you will all understand how difficult it is to deal with the anguish that you know your children are feeling.

From the moment of the birth of our children, we love, protect and try to shield them from all harm, hurt and grief. We watch them grow, try to nurture them and pray that their future will be happy, carefree, the sun will always shine on them and that dark clouds will never enter their lives. From the moment you give birth to a child all mothers dream of the beautiful grandchildren who will one day climb on their knee and call them Granny."

I cannot imagine the ache of a mother or father on learning that their child has been born with a handicap, disability or life-threatening condition of any kind. Whether this knowledge becomes known at birth, emerges slowly in the growing child or becomes apparent in a little baby boy who had a long lasting wheezy chest that would not go away. Learning about it has got to be devastating. While one reads, or is told of the various emotions which follow the discovery, the fear, denial, anger, grief, and the sense of utter helplessness felt by the parents, unless you have walked in their shoes, it is truly beyond comprehension.

But as a grandmother who has experienced her child's reaction to such a situation I can speak of the pain that I felt, not for my grandson, but for my beloved child. I was angry, at whom or what I do not know, that my child was hurting. And I could do nothing to take away her pain or alleviate her fears. I could no longer kiss the hurt and make it all better as I did when my daughter was a little girl. I could not fix it. This has been very hard for me to accept. However, with time we all grow wiser and I now know and recognize my limitations. I do what I can, when I can and being part of this group is something that I can do.

I recognize the need for support for everyone involved, and as I watched all those little boys, who were strangers to each other, till they arrived at a hotel in Baltimore on a June day in 2000, gravitate towards each other as if they had been buddies all their lives, I could not help thinking of something my Mother used to say, "A stranger is just a friend you have yet to meet." So let us grandparents not be strangers to one another, let us meet as often as we can, be it on the Internet, by "snail mail", or by phone and, if at all possible, in Baltimore this October. To those who were there last time, "Looking forward to seeing you again" and to those who were not there "Hoping to meet you this year".

Till then, God bless,
Moira Masterson

P.S. Being a grandmother is completely different from being a mother.

A CHILD'S EDUCATIONAL NEEDS

(Continued from page 11)

number from the actual number of days he has been alive. Then compare that number to the number of days children of his age and grade are normally in school. Remember that children without Barth have had more opportunities to be held, talked and read to and taken places by their immediate family and extended families, because their family's attention was not distracted by such serious issues as paying attention to every subtle change in health. Parents are very observant, and it is my

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In Memory of Lattigo Cook

LATTIGO MICHAEL COOK

May 1, 2000—January 25, 2002

Lattigo, "blonde hair, blue eyes and life from God", went to Heaven on January 25, 2002. He is survived by his parents Carol and Michael, along with his two sisters, Bailee and Dakota. He had a dog named "Boo" whom he shared his beloved chips with. He loved to share his food! He learned sign language and could also speak. He signed "more music" very often. He really enjoyed Beach Boy's music, as he bounced to the beat when he heard it. Being held and danced around the room was pure satisfaction for Lattigo. Shrek was his favorite movie.

Carol wrote to me explaining that "one morning Lattigo was playing with his Christmas toys and the sun was shining in the window and his sister put her Angel wings on him. He started to take them off and I asked Lattigo if he would put them back on for a picture. He said 'yes' and proudly posed for this picture."

Lattigo influenced many lives in his short time here. There were over 400 friends at his funeral to honor him. Lattigo will be greatly missed by all of his family. Condolences may be sent to Carol & Michael Cook, 10772 E. 875 N., Walkerton, IN 46574.

A Child's Educational Needs

(Continued from page 16)

experience that "mother's know when something is "wrong". This is probably because mothers seem to spend more time with their children while the fathers work to support the family. Even if the mom was trained as a teacher, she may not be able to put her finger on the precise concern or reason for thinking there is a problem. In addition, parents who have given birth to a "preemie" are told to think of their child as having an "adjusted age" that is based on the date at which the child was expected to be delivered rather than the child's actual birth date.

Now, what soft signs should you look for educationally? Since each child is an individual, the difficulties will vary, but answering the questions that follow with reference to your child will guide you.

- At what age did your son begin talking? What's normal? Normally developing children have a few words by 13 months, fifty words by 17 months, a few two-word combinations by 18 months and more of these by 22 months. They also have an average sentence length of 2 words at 24 months, 3.+ words at 30 months, 4+ words at 37 months and 4 1/2 words per sentence by 40 months. Children whose language is delayed (perhaps do to "adjusted"

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Remembering His Spirit

PATRICK REILLEY

December 26, 1996—October 26, 1998

Everybody was gathered in the kitchen to watch him blow out his *one* birthday candle. As the candle was lit, everyone sang: "Happy birthday to you. Happy birthday to you. Happy birthday dear Patrick. Happy birthday, to you."

We cut the birthday cake and opened the presents. We took pictures and ate ice cream. It was a special day for my little one. More special than I knew. It was to be the *only* birthday party Patrick ever had. He died just before his second year.

As Patrick's 2nd birthday drew closer, I still wanted to celebrate his life. There would be no party, no gifts, no candles on a cake, but surely this day would not go by, without *remembering,* without *honoring,* without *celebrating* the day this precious child came into my life.

We decided we would buy a gift that Patrick would have liked. We wrapped it up. We put a bow on it, and we returned to the hospital where Patrick died. We went to the Ronald McDonald House and baked lots and lots of hot cookies. We left some there and we brought some to the hospital that took such good care of our boy.

Then we took Patrick's gift to the Atrium, a large indoor play area. This is no regular playground. You won't see children running around in here. You won't hear them yelling or making noise. This is a very special play area for seriously ill children to come and get away from the routine of hospital life, for just a little while. It is a quiet, peaceful place. A place where you can see and feel the warmth of the sun. As the gift was being opened, I took a familiar look around, remembering all the happy hours Patrick and I spent here, looking at all the colorful toys.

We gave our gift to the director, to open. Yes, it was hard. Yes, I *tried* not to cry. *Yes*, I cried, but just a little. It was a healing experience for my whole family. Today was Patrick's birthday. Today his brother and sisters, his Mommy and his Daddy would remember the day he was born. It will always be a day we celebrate, with joy, and in gratitude to God, for the precious gift Patrick was to us.

In loving memory of Patrick,
His Mommy, Deanna-Lyn Reilley

(Continued from page 17) **A CHILD'S EDUCATIONAL NEEDS**

birth dates) often wait until 27 months before their first words appear. By 38 months they will have a fifty word vocabulary. Two-word combinations do not appear until 40 months with more of these predominating at 48 months. Mean sentence length at 52 months is two words, three word sentences at 63 months, four word sentences around 73 months, and 4 1/2 word sentences around 79 months. (Exceptional Learners, Hallahan and Kauffman, 2000 Allyn and Bacon publishers)

- How easily could you understand his speech? How easy could others (outside your immediate family) understand his speech?
- Is he able to identify the letters of the alphabet (both upper case and lower case or just one of these? Which ones does he NOT know?
- Can he tell the difference between the names of the letters and the sounds of the letters? Is he able to isolate and produce the sounds of each letter? Which ones does he NOT know?
- Does he have at least 50 words that he recognizes by sight? Words included in this list would be "the, and, color, look, I, you, we, mom, dad, said. his own name (first and last) one, two, three (etc). Can he blend sounds of letters into words (can, help, bed, bring)?
- Can he write his own name? Can he write his first, middle and last name?
- How does he hold a pencil? Does he hold it like most, or does he need to have more of his hand touching the pencil?
- How long does it take him to complete any of the above tasks in comparison to age or grade mates (or other children in your family or children of your friends?
- If he is in school, are his papers legible? Can he write "on the lines"? Does he leave an appropriate amount of space between letters? Does he write in upper or lower case letter? Does he mix some upper and lower case letters inappropriately when spelling words? Does he shift between upper and lower case letters? Does he make upper case letters taller than or the same size as lower case letters? Can he line up numbers for adding, subtracting, multiplying and dividing?
- Is he old enough to learn how to type? Consider using the Diana King method of teaching him to type based on a child's ability to say the alphabet in order. It is available from Educators Publishing Service. (www.epsbooks.com)
- Can he put alphabet letters in order? Can he say the letters of the alphabet in order or does he get stuck and have to go back to the beginning and start over if he gets stuck?
- How long is he able to work on an academic task (reading, writing, math) without asking to quit or putting his head down or saying he is tired?
- How many days can he work on academic tasks (either at home or school) without becoming tired?
- Is he able to stay in school all day, or does half a day (or less) wear him out?
- Have you had him tested by the local school district to determine his eligibility for special services such as identification as being other health impaired or having a learning disability?
- Does your state or local school district provide "home-bound" teachers to assist with his education?
- Do you think he would do better working with a tutor at home? Do you have a college/university near you that you might go to for assistance in finding a skilled graduate or undergraduate student who could assist? Is there a local retired teacher's organization that might assist you in finding someone to help your child at home?
- Have you filed his school papers in chronological order, particularly the ones demonstrating his difficulties?
- Is he able to read his textbooks and worksheets independently?
- Does he seem to understand something when it is initially explained, but forget it by the time he needs to do his homework?
- Does he remember what he hears better than what he reads? In other words, is his oral comprehension better than his reading comprehension? I so, he might profit from using textbooks on tape from Recordings for the Blind and Dyslexic.

If you have specific questions about any of these suggestions, please feel free to e-mail us at jstoner@navix.net.

BARTH SYNDROME IN EUROPE

Professor Peter Barth and Joke van Loo of the Netherlands are building an informative European website about Barth syndrome: You may find this website at: www.wapenveld.com/BarthSyndroom

On this site one can find information about Barth Syndrome in various European languages such as Dutch, German, Danish and French. For each Western-European country some useful links and addresses are given. Tips and advise from families or specialists in Western-Europe for this site are welcome and may be sent to: jokevanloo@hotmail.com

Our next step will be to establish a relationship with the European Organization for Rare Disorders (Eurordis), the European Neuro-Muscular Center and European associations for mitochondrial diseases, myopathies and congenital heart diseases regarding Barth syndrome and to inform them of both the European and American websites. We are fortunate to have Prof. R.J.A. Wanders, Dr. R. Duran and Dr. F. Valianpour doing research about cardiolipin and Barth syndrome at the University of Amsterdam. *-Joke van Loo*

THE POWER OF KINDNESS…..

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The Heart of the Matter…

A special place for our young men and children

We are all aware of the many difficult challenges that Barth syndrome instills upon each and every one of us. Will McCurdy, who has Barth syndrome and is now 16 years old, sent in the poem "Don't Q uit". Will writes " I have had this poem by my bed for a number of years. I look at it often and hope that others find it as helpful as I do." Also submitted is a very touching letter from a young boy who made a commitment to make a difference in the lives of these boys affected by Barth syndrome.

DON'T QUIT

When things go wrong, as they sometimes will,
When the road you're trudging seems all up hill,
When the funds are low and the debts are high,
And you want to smile, but you have to sigh,
When care is pressing you down a bit –
Rest if you must, but don't you quit.

Life is queer with its twists and turns,
As every one of us sometimes learns,
And many a fellow turns about,
When he might have won had he stuck it out.
Don't give up though the pace seems slow –
You may succeed with another blow.
Often the goal is nearer than
It seems to a faint and faltering man;
Often the struggler has given up
When he might have captured the victor's cup;
And he learned too late when the night came down,
How close he was to the golden crown.

Success is failure turned inside out –
The silver tint of the cloud of doubt,
And you never can tell how close you are,
It may be near when it seems afar;
So stick to the fight when you're hardest hit, -
It's when things seem worst that you mustn't quit.

Author Unknown

"My name is Tommy. I am 9 years old. My cousin Lattigo Cook has Barth syndrome. He died January 25, 2002. I miss him very much. In October my Aunt Cherie Schrader ran the Chicago Marathon to raise money for Barth syndrome. I wanted to help so we ordered See's Candy Bars and I sold them at soccer, at my post office and at the marathon. Please accept this $200.00. Please help the other boys.

Sincerely,
Tommy

Tommy's dad stated that Tommy raised this money while Lattigo was still with us. Tommy, bless your heart. On behalf of the Barth Syndrome Foundation, thank you so much for all of your efforts to make a difference in the lives of these boys.

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

Barth Syndrome Foundation®

The Barth Syndrome Foundation, Inc.
www.barthsyndrome.org

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