Barth Syndrome Foundation
www.barthsyndrome.org

Barth Syndrome Journal

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Saving lives through education, advances in treatment, and finding a cure for Barth syndrome.

Exciting Changes to Barth Syndrome Foundation's Research Grant Program

By Matthew J. Toth, PhD, Science Director, Barth Syndrome Foundation

Inside this Issue

PUTTING PATIENTS FIRST
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The Barth Syndrome Foundation, Inc. (BSF) and its international affiliates are pleased to announce the availability of funding for basic science and clinical research on the natural history, biochemical basis, and treatment of Barth syndrome. **Starting in 2013**, there will be two types of grant awards: **IDEA** grants for 1-2 years and **DEVELOPMENT** grants for 2-3 years with budgetary maximums of US $50,000 or $100,000, respectively over the full period. BSF's Research Grant Program allows young, non-tenured investigators to include in their submitted budget up to 75% of the direct costs amount as PI salary (10% for established investigators). In addition, for those clinical applications where volunteers must travel to a clinical research site, these travel expenses will be handled separately and will not be included in the application budget limitation. We encourage all investigators at every professional level to submit their best ideas for advancing the state of knowledge about Barth syndrome so that progress can be made in finding a specific treatment or a cure for this unusual mitochondrial disease. There are no geographical limitations to this funding.

(Cont'd on page 4)

Brayden (age 3) and Dr. Matt Toth 2013

Barth Syndrome Foundation Awards Nine Research Grants for 2012 Cycle

By Matthew J. Toth, PhD, Science Director, Barth Syndrome Foundation

The 2012 Barth Syndrome Foundation (BSF) Research Grant Program awarded nine research grants for a total of US $354,250 — the largest such commitment of BSF towards this important program. This brings the cumulative total of almost US $2.7 million in 72 grants awarded to 43 researchers since the 2002 cycle when BSF's Research Grant Program began. The quantity, quality, and sophistication of the applications for each succeeding cycle of the Research Grant Program continue to increase, and especially for 2012. Many of the applications now incorporate the use of the knockdown mouse model of Barth syndrome that BSF had wisely furnished to the scientific community only a few years ago. The maturity of the scientific and clinically-orientated applications is now providing testable ideas to consider for therapy. The 2012 awardees are listed on page 5. The scientific abstracts that the grant recipients wrote in their applications are available on BSF's website at www.barthsyndrome.org.

(Cont'd on page 5)

Barth Syndrome Foundation's Updated Roadmap for the Future

By Marc Sernel, BSF Board Chairman, and Lindsay Groff, BSF Executive Director

The Barth Syndrome Foundation (BSF) is not an organization that rests on its accomplishments. With so much at stake, we simply do not have time or money to waste. While BSF has achieved many great things since its founding thirteen years ago, we also know there is still much work to be done to reach our ultimate goal: *A world in which Barth syndrome no longer causes suffering or loss of life.* Vital to achieving any goal is having a plan to get there. As such, we set about to formulate an updated, detailed plan to chart our course over the next several years.

Many months of information gathering and strategic discussions led to the development of a detailed set of short-term objectives for the organization, as well as specific plans about how we intend to achieve these objectives. As many of you will remember, the process started by seeking input from the BSF community at large. We surveyed affected individuals, families, researchers, clinicians, and donors to learn more about various perspectives on the current state of the organization and ideas for the future. This input provided the fuel for vigorous discussion during a two-day strategic planning retreat in January, when a diverse group — including BSF staff, board members, volunteers, and international affiliates — engaged in passionate conversations, brainstorming, and focused small group sessions. The group dreamed big while also focusing on the small details.

Working groups at BSF's strategic planning meeting held in January 2013.

(Cont'd from page 2)

As the process moved forward, three overriding priorities emerged as the backbone of our plan for the organization: (1) Expand BSF Science & Medicine program to further encourage finding treatments or a cure; (2) Enhance the capacity of BSF to better meet its mission; and (3) Build confidence and trust in the BSF community and resources. Under each of these priorities, the group identified various objectives that the organization will seek to achieve in the next three years. As an example, the organization has set the ambitious goal of initiating its first clinical trial of a potential treatment for Barth syndrome in the next year. We also have decided to revamp our website, as it is our "face" to the world and our most important communication and awareness vehicle. In addition to creating a detailed list of objectives, the group also dug down one more level, discussing and deciding how to allocate the precious resources — staff time, funding, collaborative partners, etc. — that will be needed to attain each of these objectives. Since much of what we want to accomplish will require money, we also set forth ambitious plans to expand the scope and size of our fundraising efforts.

BSF is not a large group with the luxury of boundless resources. But we have always figured out a way to "punch above our weight" and do more than many think possible. We believe we have developed a plan that is faithful to the urgency, ambition, and passion of BSF's community. And we are not done; while the plan sets forth objectives that we are already working to achieve, we will treat it as a living document that will not gather dust on the shelf. We thank you for your continued support, and look forward to keeping you informed as we make progress in accordance with our plan and toward our shared goals.

Front Row (L-R): Florence Mannes, Michaela Damin, Shelley Bowen, Kate McCurdy, Susan Osnos
2nd Row (L-R): Heller An Shapiro, Sue Wilkins, Steve McCurdy
3rd Row (L-R): John Wilkins, Lynda Sedefian, Randy Buddemeyer, Hilary Vernon
4th Row (L-R): Matt Toth, David Axelrod, Lindsay Groff, Steve Kugelmann, Marc Sernel, Susan McCormack

(Photos on pages 2-3 are courtesy of BSF. 2013)

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Exciting Changes to Barth Syndrome Foundation's Research Grant Program

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Background

Barth syndrome (BTHS) is a serious X-linked genetic condition associated with cardiomyopathy, neutropenia, skeletal muscle weakness, exercise intolerance, growth delay, and diverse biochemical abnormalities (including defects in mitochondrial metabolism and phospholipid biosynthesis). Because many clinical and biochemical abnormalities of Barth syndrome remain poorly understood, we are seeking proposals for both basic science and clinical research that may shed light on any aspect of the syndrome, with the ultimate objective of developing a specific treatment or a cure.

Types of Proposals Sought

We are interested in providing financial assistance to investigators interested in exploring the field of BTHS science and/or clinical research. We anticipate that these funds might be useful as "seed grants" for the testing of initial hypotheses and the collection of preliminary data that can lead to successful long-term funding by the National Institutes of Health (NIH) and other major granting institutions around the world. In addition to those having prior research experience with BTHS, we encourage young investigators and experienced investigators that are new to the field of BTHS to submit proposals for funding.

Process

We have a competitive grant process. Applications should be of 10–15 pages in length and must follow the instructions listed on the BSF website. In general terms, detailed information about the specific aims, significance, research design and methods, personnel, facilities, and budget will be required. A one-page, "**Letter of Intent**" is required for **DEVELOPMENT** grant applicants with a due date of September 1, 2013. The "**Letter of Intent**" is optional for **IDEA** grant applicants. We strongly encourage the submission of letters of intent before the due date to allow ample time for review and feedback.

Completed applications (and/or "Letters of Intent") will be forwarded to BSF's Scientific and Medical Advisory Board (as well as to expert outside reviewers) for confidential evaluation. Response to the "Letters of Intent" will be communicated within two weeks of receipt. Based on the recommendations of the BSF Scientific and Medical Advisory Board, the BSF Board of Directors will make the final funding decisions about the grant applications. Please review our "Grants Awarded" webpage for a listing of grants that BSF and its affiliates have awarded to date.

Funding

We anticipate awarding several **IDEA** and **DEVELOPMENT** grants each year. Funds will be available soon after the successful grant applicants have been notified in early March, 2014.

Deadline

The deadline for submission of the completed research grant application is **October 31, 2013**, and grants will be awarded in early March, 2014. The deadline for the one-page "Letter of Intent", if applicable, is **September 1, 2013**.

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"In this tight funding climate, the Barth Syndrome Foundation has allowed our work, and that of many others, to continue. The Foundation helps to fund, bring together, coordinate, and disseminate research that is truly translational." *~ Colin Phoon, MPhil, MD, Associate Professor, New York University Medical Center, New York, NY*

BSF Awards Nine Research Grants for 2012 Cycle

(Cont'd from page 1)

William T. Pu, MD, PhD, Associate Professor, Boston Children's Hospital, Boston, MA
Maturation of Barth syndrome models for clinical translation
Award — US $40,000 over 1-year period
**Funding for this award was provided by Barth Syndrome Trust (UK & Europe)*

Using induced pluripotent stem cells (iPS) for defining the importance of the different mRNA isoforms of the *tafazzin* gene, for the screening of potential therapeutic compounds, and the use of the knockdown mouse model of Barth syndrome for testing the metabolic implications of the arginine deficiency found in Barth syndrome.

Colin Phoon, MPhil, MD, Associate Professor, New York University Medical Center, New York, NY
Role of mitochondria during myocardial morphogenesis in Barth syndrome
Award — US $40,000 over 1-year period
**Funding for this award was provided by BSF's Paula and Woody Varner Fund*

Studying reactive oxygen species (ROS) and how it is involved in the hypertrabeculation-noncompaction (HT-NC) trait of both the knockdown mouse model and Barth syndrome individuals.

Yuguang (Roger) Shi, PhD, Professor, Pennsylvania State University School of Medicine, Hershey, PA
Regulation of cardiomyopathy by ALCAT1 in Barth syndrome
Award — US $40,000 over 1-year period

Testing whether inhibition of the *ALCAT1* gene, a gene that also affects cardiolipin, can reverse the cardiomyopathy of the *tafazzin* knockdown mouse model of Barth syndrome.

Adam Chicco, PhD, Assistant Professor, Colorado State University, Fort Collins, CO
Mechanisms of substrate-specific impairment of oxidative phosphorylation in *taz*-deficient cardiac mitochondria
Award — US $40,000 over 1-year period

To study the knockdown mouse model of Barth syndrome which revealed a vitamin B5 deficiency and intestinal cellular lesions and that may suggest new therapeutic options for treatment.

(Photos courtesy of individual researchers.)

Angela Corcelli, PhD, Associate Professor, University of Bari, Aldo Moro, Bari, Italy
Determination of the monolysocardiolipin/cardiolipin (MLCL/CL) ratio in intact nucleated cells: A new tool for the screening of Barth syndrome
Award — US $40,000 over 2-year period
**Funding for this award was provided by the Association Barth France*

Developing a quick, cardiolipin assay using MALDI-TOF mass spectrometry analysis using small blood samples without prior purification.

Richard Epand, PhD, Professor, McMaster University, Hamilton, Ontario, Canada
Relationship between membrane physical properties and the action of *tafazzin*
Award — US $40,000 over 2-year period
**Partial funding for this award was provided by Barth Syndrome Foundation of Canada*

Using nuclear magnetic resonance (NMR) studies of mitochondrial membranes altered by *tafazzin* dysfunction to explore the unique curvature (non-bilayer) attributes of cardiolipin.

Matthew P. Gillum, PhD, Research Assistant Professor, University of Iowa, Iowa City, IA
Implications of phosphatidylserine deficiency in skeletal muscle and heart of ROSA26-*taz* shRNATet-on mouse model of Barth syndrome
Award — US $40,000 over 1-year period

Investigating the phosphatidylserine (PS) deficiency in the knockdown mouse model of Barth syndrome and its effects on programmed cellular death (apoptosis).

Robert Ryan, PhD, Senior Scientist, Children's Hospital and Research Center at Oakland, Oakland, CA
Cardiolipin replacement therapy for Barth syndrome
Award — US $40,000 over 1-year period

Testing the potential of lipid replacement therapy (nanodisks of protein and cardiolipin) for the treatment of Barth syndrome.

Michael T. Chin, MD, PhD, Associate Professor, University of Washington, Seattle, WA
***Tafazzin* enzyme replacement therapy for heart muscle in Barth syndrome**
Award — US $40,000 over 1-year period

Testing whether enzyme replacement therapy is possible for the treatment of Barth syndrome.

Barth Syndrome Foundation Files With the FDA for Orphan Drug Designation with Bezafibrate

By Matthew J. Toth, PhD, Science Director, Barth Syndrome Foundation

Over the last several months, a team of Barth syndrome researchers (many of them members of BSF's Scientific & Medical Advisory Board) has been working with the SMARTT (Science Moving TowArds Research Translation and Therapy) group at the National Heart Lung and Blood Institute of the National Institutes of Health (NIH) to prepare for testing the pharmaceutical called bezafibrate in the treatment of Barth syndrome individuals. Encouraged by the work done by Mindong Ren, PhD and Colin Phoon, MPhil, MD, the bezafibrate team recently filed for Orphan Drug Designation with the US Food and Drug Administration (FDA). Bezafibrate is a pharmaceutical that has been used for over 25 years to treat high triglyceride levels — a risk factor for coronary heart disease like high cholesterol. The team looks upon bezafibrate as a potential pharmaceutical treatment because of the results in the Ren and Phoon labs, and its long and safe use in Europe, the UK, and Canada. One step in this process is to file with the FDA to use this drug for a rare disease — Orphan Drug Designation. This application, if granted, provides BSF with important communication avenues with the FDA who will ultimately need to agree to the testing of this drug and if successful, ultimately approve this drug for use in Barth syndrome. Orphan Drug Designation allows the Barth Syndrome Foundation (BSF) to take advantage of certain NIH and FDA funding programs to aid in the development of this drug as a treatment. BSF filed for Orphan Drug Designation on May 8, 2013. There will be a long road ahead with this, as any project for the approval of the use of a drug in humans is an involved, lengthy and thorough process, but this is a very exciting and critical early step forward.

Barth Syndrome Foundation Facilitates Truly Translational Research

By Colin Phoon, MPhil, MD, Associate Professor, New York University Medical Center, New York, NY

" In ongoing work, my eyes are now opened to the broader importance of cardiolipin in human health and disease, including common diseases such as heart failure, diabetes, and aging. But the focus remains always on our "little corner of the world" that is the Barth syndrome community." ~ *Colin Phoon, MPhil, MD*

Dr. Colin Phoon
(Photo courtesy of Dr. Phoon [2013])

My parents raised me to make our own little corner of the world a better place. I came to medicine focused on individual patient care, teaching at the grass-roots level, and modest, but not ambitious, scientific discovery. Still, I love learning new things, and working at an academic medical center leads to many chances to work with smart, creative people. And, so when Michael Schlame approached me one day about collaborating on a mouse model of Barth syndrome — at the time, we were both in the OR with a child undergoing heart surgery — I jumped at the opportunity. This has turned out to be one of the most fortuitous collaborations in my career. I had only a general knowledge of Barth syndrome, and had no idea about the team I was about to join — or the community I would be introduced to. What better way to learn new things than to take on a whole new field, mitochondrial biology?

I am blessed to have such outstanding close collaborators as Mindong Ren, Michael Schlame, and David Stokes, but also others from the Barth syndrome community including old friends such as Barry Byrne. For me, everything came together at the Barth Syndrome Foundation's 2012 International Scientific, Medical, and Family Conference. Here, I got to see a broad swath of the science of Barth syndrome, meet like-minded scientists and doctors, and most importantly, meet the Barth families. In this tight funding climate, the Barth Syndrome Foundation has allowed our work, and that of many others, to continue. The Foundation helps to fund, bring together, coordinate, and disseminate research that is truly translational: How else have I been able to rub elbows with yeast researchers, work on membrane biochemistry, treat mice (and men!), and collaborate on a clinical trial?

The most important questions in my career arose from our initial findings in this mouse model: What is the role of mitochondria during heart development, and how does this role change once the heart matures? In ongoing work, my eyes are now opened to the broader importance of cardiolipin in human health and disease, including common diseases such as heart failure, diabetes, and aging. But the focus remains always on our "little corner of the world" that is the Barth syndrome community.

Barth Syndrome Researcher Wins Prestigious E. Mead Johnson Award For Outstanding Pediatric Research

Dr. William Pu
(Photo courtesy of BSF [2012])

William Pu, Associate Professor, Harvard Medical School
Department of Cardiology, Boston Children's Hospital, Boston, MA
Nominated by David Clapham

Modeling cardiomyopathy using human induced pluripotent stem cells

BSF congratulates Dr. William Pu on receipt of the **E. Mead Johnson Award for Research in Pediatrics** at the 2013 annual meeting of the Pediatric Academic Societies. This prestigious award honors outstanding clinical and laboratory research achievements in pediatrics. Dr. Pu presented a talk at the meeting titled "**Modeling cardiomyopathy using human induced pluripotent stem cells**".

Dr. Pu has advanced the understanding of mechanisms that regulate heart development and adult heart function. His work has revealed transcription factors and transcriptional regulatory mechanisms that control heart morphogenesis and the stress response of the post-natal heart. Dr. Pu's research has also highlighted the contribution of distinct cell types to formation, vascularization, and injury responses of the heart. Most recently, Dr. Pu's research has used insights from heart development to uncover new potential approaches to improve heart repair and regeneration.

Dr. Pu was also awarded a BSF Research Grant Award titled, Maturation of Barth syndrome models for clinical translation (Award — US $40,000 for over 1-year period). Funding for this award was provided by Barth Syndrome Trust (UK & Europe). Please visit BSF's website to read the abstract.

Barth Syndrome on Capitol Hill!

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

The voice of the Barth syndrome community was amplified through the American Heart Association's (AHA) You're the Cure on the Hill. In April, I attended a two-day event which included advocacy training, a rally for medical research, and meetings with U.S. congressional representatives. Over 300 advocates united to request the restoration of funding to the National Institutes of Health (NIH), the largest medical research funding agency in the world. The U.S. federal budget cuts that took effect earlier this year eliminated $1.5 billion from the NIH budget. These cuts are already causing the termination or delay of promising research projects throughout the country. The Barth Syndrome Foundation joined AHA, along with other groups in the medical research community, to urge Congress to restore these funds. Thank you, AHA for allowing many hearts to speak with one voice.

Lindsay Groff attends Rally for Medical Research.
(Photo courtesy of BSF [2013])

Fellow advocates at You're the Cure on the Hill.
(Photo courtesy of BSF [2013])

Awareness of Barth Syndrome Continues to Grow

There has been a significant increase in Barth syndrome (BTHS) related peer-reviewed journal articles published. To date, a total of **71** articles have been published on BTHS research conducted with the support of BSF and/or BSF affiliate funding (denoted below with †**)** and publications that acknowledge biological samples and/or information from Barth families, the Barth Syndrome Registry and Repository, and/or BSF affiliates (denoted below with ▼). Listed below are articles relevant to BTHS that have been added to BSF's library since the last issue of the Barth Syndrome Journal. To view the complete bibliography on BTHS, please visit www.barthsyndrome.org.

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Funding Opportunities Relevant to Barth Syndrome Research

The following ongoing research initiatives at organizations other than BSF are particularly relevant to Barth syndrome:

National Institutes of Health (NIH)

Improvement of Animal Models for Stem Cell-Based Regenerative Medicine (R01) Funding Opportunity Announcement (FOA) Number: PAR-13-114	Open Date: May 5, 2013 Application Due Date(s): Standard dates apply, by 5:00 PM local time of applicant organization Expiration Date: May 8, 2016 http://grants.nih.gov/grants/guide/pa-files/PAR-13-114.html
Purpose: This FOA encourages Research Project Grant (R01) applications from institutions and organizations proposing research aimed at characterizing animal stem cells and improving existing, and creating new, animal models for human disease conditions. The intent of this initiative is to facilitate the use of stem cell-based therapies for regenerative medicine. The initiative focuses on the following areas: (1) comparative analysis of animal and human stem cells to provide information for selection of the most predictive and informative model systems; (2) development of new technologies for stem cell characterization and transplantation; and (3) improvement of animal disease models for stem cell-based therapeutic applications.	

Differentiation and Integration of Stem Cells (Embryonic and Induced-Pluripotent) Into Developing or Damaged Tissues (R01) Funding Opportunity Announcement (FOA) Number: PAR-13-094	Open Date (Earliest Submission Date): May 5, 2013 Letter of Intent Due Date(s): 30 days before application due date Expiration Date: September 8, 2016 http://grants.nih.gov/grants/guide/pa-files/PAR-13-094.html
Purpose: The primary focus of the FOA is to promote in vivo studies of stem cells in animal models and in humans (if applicable) to better understand how stem cells function within developing or damaged tissues. The areas of emphasis would include systematically profiling and cataloging changes at genetic and epigenetic levels that take place in stem cells and their microenvironment. The purpose is to gain in-depth knowledge of the mechanisms involved in: progressive differentiation of Embryonic Stem Cells (ESCs) into embryonic lineages, progenitor cells and specialized cell types; adult stem cells/progenitor cells during tissue regeneration and wound healing; and Induced Pluripotent Stem Cells (iPSCs) at the site of injury during stem cell therapy.	

Differentiation and Integration of Stem Cells (Embryonic and Induced-Pluripotent) Into Developing or Damaged Tissues (R21) Funding Opportunity Announcement (FOA) Number: PAR-13-095	Open Date (Earliest Submission Date): May 16, 2013 Letter of Intent Due Date(s): 30 days before application due date Expiration Date September 8, 2016 http://grants.nih.gov/grants/guide/pa-files/PAR-13-095.html
Purpose: This funding opportunity is intended to encourage innovative and high risk/impact research in the area of stem cell biology, to be explored in model organisms. The research proposed under this program can explore approaches and concepts new to this area; development of new technologies; or initial research and development of data upon which significant future research may be built. The primary focus of the FOA is to promote in vivo studies of stem cells in animal models and in humans (if applicable) to better understand how stem cells function within developing or damaged tissues. The areas of emphasis would include systematically profiling and cataloging changes at genetic and epigenetic levels that take place in stem cells and their microenvironment. The purpose is to gain in-depth knowledge of the mechanisms involved in: progressive differentiation of Embryonic Stem Cells (ESCs) into embryonic lineages, progenitor cells and specialized cell types; adult stem cells/progenitor cells during tissue regeneration and wound healing; and Induced Pluripotent Stem Cells (iPSCs) at the site of injury during stem cell therapy.	

Discovery of Genetic Basis of Mendelian or Monogenic Heart, Lung, and Blood Disorders (X01) Funding Opportunity Announcement (FOA) Number: PAR-11-307	Open Date (Earliest Submission Date): September 18, 2011 Letter of Intent Due Date: September 19, 2011; April 16, 2012; April 15, 2013; and April 14, 2014 Application Due Date(s): October 18, 2011; May 14, 2012; May 14, 2013; and May 14, 2014 Expiration Date: May 15, 2014 http://grants.nih.gov/grants/guide/pa-files/PAR-11-307.html
Purpose: To stimulate discoveries of the genetic basis of Mendelian or monogenic disorders that significantly affect heart, lung, and blood (HLB) systems, the NHLBI invites X01 to use the genome-wide sequencing capacity of the Mendelian Disorders Genome Centers which are funded under the HG-10-016.	

(Cont'd on page 10)

Funding Opportunities Relevant to Barth Syndrome Research

(Cont'd from page 9)

National Institutes of Health (NIH)

Health Promotion for Children With Physical Disabilities Through Physical Activity and Diet: Developing An Evidence Base (R21) Funding Opportunity Announcement (FOA) Number: PAR-11-284	Open Date (Earliest Submission Date): September 16, 2011 Letter of Intent Due Date: 30 days prior to applicable receipt date Expiration Date: September 8, 2014 http://www.grants.gov/search/search.o?mode=VIEW&oppId=110713

Purpose: This Funding Opportunity Announcement (FOA) encourages Exploratory/Developmental Research Grant (R21) applications that will improve our understanding of how patterns of physical activity and dietary choice affect the health and fitness of children with physical disabilities. Proposed research should account for the functional limitations of children with disabilities and their nutritional needs, as well as the physiological, psychosocial, and environmental factors that play a role in determining the health of this population.

Health Promotion for Children With Physical Disabilities Through Physical Activity and Diet: Developing An Evidence Base (R01) Funding Opportunity Announcement (FOA) Number: PAR-11-288	Open Date (Earliest Submission Date): September 5, 2011 Letter of Intent Due Date: 30 days prior to applicable receipt date Expiration Date: September 8, 2014 http://grants.nih.gov/grants/guide/pa-files/PAR-11-288.html

Purpose: This Funding Opportunity Announcement (FOA) encourages Research Project Grant (R01) applications that will improve our understanding of how patterns of physical activity and dietary choice affect the health and fitness of children with physical disabilities. Proposed research should account for the functional limitations of children with disabilities and their nutritional needs, as well as the physiological, psychosocial, and environmental factors that play a role in determining the health of this population.

Innovative Therapies and Tools for Screenable Disorders in Newborns (R01) Program Announcement (PA) Number: PAR-10-230	Opening Date: September 5, 2010 Letters of Intent Receipt Date: 30 days prior to application due date Application Due Date: See http://grants1.nih.gov/grants/funding/submissionschedule.htm Expiration Date: September 8, 2013 http://grants.nih.gov/grants/guide/pa-files/PAR-10-230.html

Purpose: This FOA, issued by the Eunice Kennedy Shriver National Institute of Child Health and Human Development, the National Institute of Diabetes and Digestive and Kidney Disease, the National Institute of Neurological Disorders and Stroke, and the National Institute on Deafness and Other Communication Disorders encourages Research Project Grant applications from institutions/ organizations that propose research relevant to the basic understanding and development of therapeutic interventions for currently screened conditions and "high priority" genetic conditions for which screening could be possible in the near future. In this FOA, a "high priority" condition is one for which the development of an efficacious therapy would make the condition amenable to newborn screening.

American Society of Hematology
Patient Group Research Grant Opportunities

To draw together the multitude of hematology-related research grant opportunities that are available through patient groups, the Society has created a section on the ASH Web site that simplifies your search for requests for blood and blood disease research topics. (http://www.hematology.org/Research/2874.aspx

Children's Cardiomyopathy Foundation

The Children´s Cardiomyopathy Foundation (CCF) offers two annual grant programs to support innovative basic, clinical, population, or translational studies relevant to the cause, diagnosis, or treatment of cardiomyopathy (dilated, hypertrophic, restrictive, left ventricular non-compaction, or arrhythmogenic right ventricular cardiomyopathy) in children under the age of 18 years. The goal of CCF´s grant programs is to advance medical knowledge of the basic mechanism of the disease and to develop more accurate diagnostic methods and improved therapies for children affected by cardiomyopathy. (http://www.childrenscardiomyopathy.org/site/grants.php)

United Mitochondrial Disease Foundation

The United Mitochondrial Disease Foundation (UMDF) Research Grant Program began in 1997 out of a desire to fund research toward diagnoses, treatments, and cures for mitochondrial disease. (http://www.umdf.org/site/c.dnJEKLNqFoG/b.3790285/k.6CE6/Research_Grant_Program.htm)

SAVE THE DATE!

7th International Scientific, Medical & Family Conference
June 23-28, 2014

Hilton Clearwater Beach Resort, Clearwater, Florida, USA

"…I am not aware of any other patient advocacy group that hosts simultaneous educational sessions for specific audiences such as this. I was even surprised when I learned BSF also leverages this occasion to conduct clinical research to advance the knowledge about the disease. The exceptional quality of your educational programs was made clear to me from the conversations I shared with the scientific and medical professionals attending this event." *~ Marion Burton, MD (President of the American Academy of Pediatrics)*

Save the date! The 2014 Barth Syndrome International Scientific, Medical & Family Conference is scheduled for June 23-28, 2014 at the Hilton Clearwater Beach Resort located in Clearwater, Florida. The hotel is right on the beach with two pools, tons of restaurants, ample shopping, and exciting activities all within walking distance.

Call for Poster Abstracts

The Barth Syndrome Foundation 2014 Scientific and Medical Conference Organizing Committee (COC), comprised of members of the Barth Syndrome Foundation international Scientific & Medical Advisory Board, invites the submission of abstracts for poster presentations related to the scientific and/or clinical aspects of Barth syndrome. **The deadline for abstract submission is April 15, 2014.** All submitted abstracts will be peer-reviewed by the COC. Once accepted, the submitting author will be expected to present his/her corresponding poster at a specific time during the Conference.

All Conference registrants are encouraged to submit abstract(s)/poster(s) of their work. Poster presenters are also encouraged to apply for a stipend to help defray the cost of their attendance. Program and application information will be available at www.barthsyndrome.org.

Scholarship Program

The Barth Syndrome Foundation offers a limited number of travel scholarships for qualifying physicians, clinical residents/fellows/students, nurses, and other allied health professionals to help defray the cost of attending the 2014 Conference. This program is designed to encouraged medical practitioners to increase their knowledge about and improve their care of Barth syndrome individuals. Program and application information will be available at www.barthsyndrome.org.

Barth Syndrome Clinic at Kennedy Krieger Institute

Another clinic for Barth syndrome was held at Kennedy Krieger Institute on Tuesday, June 4, 2013. The Barth Syndrome Clinic at Kennedy Krieger Institute is an interdisciplinary clinic dedicated to the diagnosis and treatment of Barth Syndrome. It provides expert care for children and adults who have, or are suspected of having, Barth syndrome. This time, four boys were seen by a number of specialists during back-to-back appointments. The day prior to the clinic, families and clinicians were interviewed for videos that will be used to increase awareness and solicit donations. Once again, the Woodwards arranged a group dinner for the families and clinicians to socialize in a relaxed setting.

Ruth Penn, R.D.C.S. performing echo at clinic.

Dr. Richard Kelley consults with family at clinic.

Families gather in Baltimore to attend the clinic. *(Photos courtesy of BSF 2013)*

Video Win is Worth a Million Words!

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

Nicole and Devin (age 8)

Kevin (age 24)

We've won, and if a picture says a thousands words, then a video speaks a million!! Thanks to Parent Advocate Kristi Pena of Mississippi, Barth Syndrome Foundation (BSF) won a professionally produced, directed, shot, and edited video to raise awareness. Over twenty charities competed in the Birds Nest Foundation's contest. Submissions took place on Pinterest where entrants pinned photos that best communicated the charity's mission. Four finalists were chosen by Birds Nest Foundation including: Stomp Out Bullying, Jumpstart, Much Love Animal Rescue, and Barth Syndrome Foundation. Birds Nest Foundation posted information about each charity on its Facebook page, and the charity with the most "likes" was named the winner.

Avis Richards, the founder and CEO of the Birds Nest Foundation said, "The Barth Syndrome Foundation's work in focusing resources on a little known, under-diagnosed disease is a perfect example of a non-profit that badly needs a voice in promoting their cause. We, at Birds Nest Foundation, are thrilled to provide that voice and excited to begin production. We're also very thankful to all the charities that didn't win, and we believe that just participating in the contest helped our finalists spread awareness. I'm very optimistic by this new era of social media. It's allowing charities like the Barth Syndrome Foundation to reach other affected families, find new sources of promotion, and forge cross-sector partnerships with organizations like Birds Nest Foundation."

Birds Nest Foundation/Productions is a 501(c)3 non-profit creative group that travels the world to provide media content for non-profits, charities and NGOs. The award winning team of experts have produced exceptional videos for organizations associated with Goldie Hawn, John Legend, Michael Bolton, Andre Agassi, and Fran Drescher as well as Autism Speaks, Susan G. Komen for the Cure. BSF is proud to be added to their list of clients and will share the video on our website once it is ready.

(Photos courtesy of Birds Nest Foundation 2013)

A Better Understanding — A Fuller Life

By Jon, Affected Individual

"The Barth Syndrome Foundation's (BSF) programs offer an incredible social support network. The work of BSF is truly indispensible to all people affected by Barth syndrome, and I am personally indebted to them for their tireless efforts. Today, I am able to live a fuller life granted by a better understanding of the root cause of my complications." *~ Jon*

Jon, age 8
(Photo courtesy of Jon)

My name is Jon, and I was diagnosed with Barth syndrome at age 27. Last spring, subsequent to moderate weakening of my heart, genetic testing at Johns Hopkins Medical Center revealed that I have Barth syndrome. This revelation was not a big surprise to my family and me given my medical history.

Although my diagnosis came only when I was older than most with this syndrome, I have experienced some of the common symptoms of the disorder with fluctuating severity throughout my life. At six months old, during a routine checkup, my pediatrician detected irregular heartbeats. The physician admitted me to a local hospital where his concern was echoed by a team of cardiologists. The outlook appeared dismal. Immediately, I began an assortment of cardiac drugs and remained in the hospital for observation.

There were great doubts about the ability of the drugs to help me, and I was given only two weeks to improve. Fortunately, to the astonishment of the doctors, my heart began to strengthen. Within two weeks I was released from the hospital with a full medicine regimen and a mysterious diagnosis of cardiomyopathy. Since then, I have largely remained in stable condition with a few intermittent cardiac procedures, medicine, and routine cardiology follow-ups.

Undeniably, health complications have had a defining impact on my life, marked by times of greater illness and certain physical limitations. Yet, this aspect remains invisible to the countless people I have interacted with at school, work, and elsewhere. What would seem irregular to most has become a normal part of me, and I have mastered the art of coping with it. I have persevered and excelled through life's stages at a rate comparable to my peers and maintain a normal life.

I am thankful for my parents' support, which allowed me to maintain a positive outlook, enjoy life, and focus on academics. While elementary school proved a bit of a challenge, I became an honor student in middle school and followed an advanced placement program in high school. I graduated with distinct honors and recognition from several academic societies and clubs.

After graduation, I attended George Mason University where I received my undergraduate degree in Finance and Economics. I was on the dean's list every semester and invited to join numerous honor societies. I graduated from college in 2010 and received the Excellence in Finance award presented to the top five students from the School of Management's graduating class. A couple of months before graduating, I was hired by a major financial institution as a financial analyst, where I have worked for the past three years. I plan to start my MBA in the next two years, with aspirations to become an entrepreneur.

While I am lucky not to have had some of the medical difficulties that others have encountered, I am proud that I have successfully assimilated into society, despite the pronounced obstacles I have faced. I do wonder, however, how things would be different for me if my diagnosis had come earlier. With recent nutritional and physical therapy, I have experienced remarkable improvement in overall stamina and in the strengthening of my heart muscle which has enhanced my overall quality of life.

In addition to the support from my parents, two other groups have helped me through the recent diagnosis. The multidisciplinary approach to understanding Barth syndrome at the clinic at Kennedy Krieger Institute has proven to be an invaluable resource. In addition, the Barth Syndrome Foundation's (BSF) programs offer an incredible social support network. The work of BSF is truly indispensible to all people affected by Barth syndrome, and I am personally indebted to them for their tireless efforts. Today, I am able to live a fuller life granted by a better understanding of the root cause of my complications.

Sharing Hope, Encouragement, Joy, and Comfort

By Jarrod Robertson, Volunteer

"I saw hope and energy focused to combat this illness. I saw eagerness and determination in mothers and fathers seeking to help researchers and clinicians find answers. I saw bravery in the face of turmoil. I saw one mother honoring her son by creating a medium through which not only information and data can be shared, but through which encouragement and hope can also be shared." *~ Jarrod Robertson, Volunteer*

Jarrod and Sue Wilkins
at BSF's 2012 Conference.

I drove alone to my first BSF conference in 2010 to volunteer in assisting Sue Wilkins in the clinical portion of the conference and to get an idea of what it was like to live with, to treat, and to research Barth syndrome. The four-hour drive to Orlando gave me a lot of time to ponder and speculate on what I was about to experience. Before this conference, I had never been exposed to anything quite like it. To be honest, I was rather apprehensive about going because I was aware of the devastating prognosis that comes with Barth syndrome. I had just completed my first year of undergraduate education in the field of Exercise Science and was only beginning the journey that has led me to where I am today.

Any shred of apprehension I had about this conference went out the window in the first few hours I spent there. With each and every interaction I had, whether it was with an affected individual or family member, or with a volunteer or clinician, I was inspired and uplifted. Before I attended the conference, I wondered what the atmosphere would be like in the face of this terrible illness. Just as I expected, I saw sadness and grief. However, I also saw something I did not expect. I saw hope and energy focused to combat this illness. I saw eagerness and determination in mothers and fathers seeking to help researchers and clinicians find answers. I saw bravery in the face of turmoil. I saw one mother honoring her son by creating a medium through which not only information and data can be shared, but through which encouragement and hope can also be shared.

My heart was set on a career in medicine long before my first experience with the BSF conference in 2010. However, what I experienced in 2010 and again in 2012 helped me mature into a more worthy candidate for a career as a physician. During these conferences, I was involved in helping collect data for the doctors and researchers by taking and recording measurements and vitals of the boys. Through my own personal interactions with the boys and their families and through observations of the interactions between the clinicians and the families, I learned a valuable lesson that will make me a better physician.

This lesson was summed up by Hippocrates in his charge to all physicians. “Cure sometimes, treat often, comfort always.” This simple quote carries tremendous weight as a reminder to all physicians that there are many ways to heal. My experience with the BSF conferences and with the families and clinicians involved helped me to see the importance of these words firsthand. The BSF conference is a truly unique time for so many wonderful things to happen. Brilliant advances in care and treatment are aided with the gathering of data for researchers, but just as importantly families, physicians, volunteers, and anyone involved can come together to share hope, encouragement, joy, and comfort. As I continue my training at The Florida State University College of Medicine to become a compassionate physician, I am thankful for my experiences with the Barth Syndrome Foundation and hope for many more in the future.

Jarrod helping at BSF's 2012 Conference.
(Photos courtesy of Jarrod Robertson & BSF 2012)

Barth Syndrome Trust
Update from the Chair...

By Michaela Damin, Chair, Barth Syndrome Trust (UK & Europe)

In May this year, I completed my term on the BSF Board, and following best practice in the US, which limits the length of service in such positions, I stepped down. I'm thrilled that Cathy Ritter from the Barth Syndrome Foundation of Canada will be joining the BSF Board. It has been a real privilege to serve our families and I would urge you all to consider a volunteering or leadership role in BSF or the affiliates. It's true that we have so little time, but, the way I see it, the rewards are beyond measure. There is no better investment of our limited spare time than in our mission to find a treatment and one day a cure for this condition which affects us all so closely. I'm not going anywhere though! I'm still hard at work here in the UK in my role as Chairperson of BST, and we have some exciting times ahead of us.

Funding research

William T. Pu, MD, Associate Professor, Children´s Hospital of Boston, Boston, MA, **Maturation of Barth syndrome models for clinical translation**. Award — US $40,000 over 1-year period

This year the Trustees of BST were thrilled to be able to approve the funding for Dr. Bill Pu's ground-breaking research. Dr. Pu has developed induced pluripotent stem cells (iPS cells) from the skin cells taken from two Barth syndrome individuals and uses them to analyze for biochemical abnormalities at the cellular level. Using state-of-the-art machines to monitor mitochondrial functions, he will explore which isoform(s) of human *tafazzin* mRNA (there are at least 6 isoforms) is able to correct these biochemical dysfunctions. He will also use these iPS cells to screen for therapeutic compounds. In addition, Dr. Pu will use potential stressors of the knockdown mouse model (arginine deficiency or 2-deoxyglucose treatment) to test whether this exacerbates the known cardiac problems of this mouse model of Barth syndrome.

Other programmes

Our Awareness Programme is another area which has been supported through your donations and fundraising efforts. Last year, our new website was launched at www.barthsyndrome.org.uk. This immediately proved its worth, attracting new members and new interest in Barth syndrome and our work.

The Family Services Programme in the UK includes the funding of special Family Days as well as financial assistance to families travelling to attend the Barth Clinics in Bristol every year.

Siblings enjoy meeting friends at the Clinic. 2012

(L-R) Mitchell and Jack 2013

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Barth Syndrome Trust
Update from the Chair...

(Cont'd from page 15)

BST Financial Summary for the 2012 period

Income	£23,629
Expenditure	£35,380
Group Development	£ 683
Fundraising and Administration	£ 350
Family Support	£ 2,671
Awareness	£ 6,881
Science and Medicine	£24,203
Other	£ 592

The Clinic

Congratulations to our brilliant Bristol Team for another successful and well-attended clinic in April. This was followed by a great day out at @Bristol, an exciting, interactive Science Museum. This was a wonderful opportunity to meet some new friends and see some old ones again. Special thanks to the staff at the museum who were incredibly friendly and helpful.

Upcoming meetings in Bristol

In July 2013, the Bristol Service Team will be hosting a special day-long meeting for families who have experienced the loss of children to Barth syndrome or who have struggled with miscarriages. The agenda for this meeting will be family-led so we urge you to please let us know what you would like to get out of this meeting.

A separate meeting will also be held at the same time for all those who have been diagnosed as carriers of the condition and who are looking for some practical advice and information surrounding possible future family planning. We look forward to seeing you there.

Save the Date

Bereavement and Carrier Meeting
Provisional date: Wednesday 17th July

Barth Syndrome Service
Bristol Royal Hospital for Children

Save the Date

Barth Syndrome Service Clinic
Bristol Royal Hospital for Children
Thursday 3rd and Friday 4th October 2013

Followed by a Family Gathering on Saturday 5th October 2013

Update from the Barth Syndrome Service Team

By Debbie Riddiford, Barth Syndrome Clinical Nurse Specialist, Bristol Royal Hospital for Children

Dedicated Experts

There have been many changes over the past year. We were delighted to welcome Dani Goodman, Occupational Therapist, Lucy Jones, Physiotherapist, and Charlotte Nicol, Clinical Nurse Specialist, to the team. They have been a fantastic help and support to the families. Dani is currently on maternity leave, but hopes to return for the clinic in October.

Charlotte is available as a contact at times during the week when Debbie is not working.

(Cont'd on page 17)

(Cont'd from page 16)

Charlotte Nicol

Introducing Charlotte Nicol — Clinical Nurse Specialist

I have worked in Bone Marrow Transplantation / Oncology / Haematology at the Bristol Royal Hospital for Children since qualifying as a Paediatric Nurse in 1997. I currently work part-time as the Paediatric BMT Clinical Nurse Specialist / Co-ordinator, a role which involves supporting families throughout the whole transplant process. This has given me plenty of experience of advising patients and their families about living with neutropenia.

I have worked with Dr. Colin Steward throughout my career and was excited when the opportunity arose to work alongside him and Debbie Riddiford in the Barth Syndrome Clinical Nurse Specialist role. It feels great to be part of such a dedicated, multi-disciplinary team and all of the members have been really welcoming, especially in teaching me so much about Barth syndrome.

It was my great pleasure to meet many of the boys, men, and their families at the recent clinic in Bristol, and I am really looking forward to working with all of you more and hopefully bringing some of my haematology knowledge and skills to the Service.

. . . and Lucy Jones, Physiotherapist

I qualified as a physiotherapist in 2008. I joined the Bristol Royal Hospital for Children in May 2012. I initially started working in the Rheumatology Department and am now working with the Oncology team. I have now also taken on the role as the physiotherapist for the Barth Syndrome Service, and am really looking forward to being part of such a specialist service and will strive to improve the care I provide. This will involve improving the links with local therapy teams as well as input to school/nursery when appropriate.

I have already learnt a huge amount from other highly specialised members of the team and am very keen to develop my knowledge further over the years to come. It was brilliant to meet so many people in the recent clinic and I am looking forward to meeting many more in the subsequent clinic in September.'

. . . and Dani Goodman, Occupational Therapist

Dani Goodman has also recently joined the Barth Syndrome Service, as a specialist Occupational Therapist. She has worked at the Children's Hospital for a number of years and has a wide range of expertise. She is particularly interested in looking at the sensory needs of people with Barth syndrome and the effect this has on them in everyday life.

Ann Exon, New Administrator

Ann Exon will join us at the end of the month. This means that there will be an extra person available to contact if any families have questions or queries.

Parent/Clinician Consultations

We have been listening to feedback from the families who have suggested that parents have some time during clinic appointments to talk to clinicians without children being present. All the members of the team are very happy to offer this service. Please talk to Debbie or Charlotte about how you would like to use your clinic appointment.

Dr. Garratt (Clinical Psychologist) is available throughout the clinic. If at any time you would like an appointment with her please let us know before you come to clinic.

Revised Leaflet for Young People

We are currently putting together some information for young people about Barth syndrome. We have had some really helpful feedback from some of the boys and their families, and are currently re-writing this leaflet. If you would like a copy of the draft leaflet please get in contact with us, we would be delighted to receive further feedback.

Feedback from Families

We are really grateful to everyone who came to the last clinic who gave us some valuable feedback about our service questionnaires. These questionnaires will help us understand how well the service is supporting young people and their families and will highlight anything that we could do differently to improve this service. We will be asking families to complete these once a year and would really appreciate your help in this. This will help us to continue to offer a service that is based around your needs and requirements and give us important information to send to commissioners to tell them about the service we are delivering. I would like to thank all of the families for participating in these assessments and agreeing to the publication of the overall data which may benefit others in the Barth syndrome community in the future.

(Cont'd on page 18)

Update from the Barth Syndrome Service Team

(Cont'd from page 17)

My Role in the National Barth Syndrome Service

By Cara Roberts-Collins

Cara Roberts-Collins
(Photo courtesy of Cara Roberts-Collins 2013)

The Barth Syndrome Trust kindly funded my neuropsychological assessment post for one year. Some parents had reported that their sons were finding it difficult to pay attention, concentrate, and learn at school. We wanted to get a better understanding of these difficulties so that we could provide additional information to support families and schools to help boys make the most of their school years.

Initially, the task of assessing 15 different families who were spread around the UK was a little daunting! However, with the excellent support of the NHS Barth Syndrome team, the Barth Syndrome Trust, and the boys' schools, I was able to plan the assessments and work closely alongside each of the families. All of the boys and their families that I have met over the past year have been so warm, friendly, and welcoming. There is such a huge amount of love and care for these boys, and I was struck by the families' positivity and determination when coping with such a challenging condition as Barth syndrome. Cara and the Bristol Service team will report on the findings of this research in the near future.

Please visit the NHS Service website at www.uhbristol.nhs.uk/barthsyndromeservice for more information.

Busy for Barth — Recent Fundraising Efforts

A big thank you to everyone who has helped us through their generosity in time or donations. Special donations were received in loving memory of Philip Brown, Isaiah Chahal-Henry, Jack Reddin, Russell Riseborough, Oscar Stobart-Hook, and Benjy Thorpe. Below are just some of the recent fundraisers in aid of Barth Syndrome Trust.

Tireless Terri's Tennis Special: Terri Allison, has over the years, raised thousands of pounds. This year Terri's Oakley Tennis Tournament raised £500 on a day of challenging tennis, scrumptious tea and cakes, and the usual good-humoured raffle.

Winner, Liam (age 13), with Organiser, Terri Allison. 2013

A close shave for Tommy. 2012

Tommy's Hair-raising Fundraising: What better way of spending Christmas 2012 on an oil rig in the cold and stormy North Sea than shaving off all your hair and well-appointed beard to raise funds? Tommy Anderson's workmates on the BP Buzzard platform, family and friends all over the world rallied round and gave £2030.

A Fond Farewell with thanks: Richard and Heather Oram raised £428 in their last annual Overton Quiz. Richard's quizzical questions have baffled the wits of the teams in the Methodist Hall for years. Heather's efficient organisation ensured the success of the evening. Many thanks Richard and Heather, and best wishes for your new venture, charity work abroad.

Busy with sales.
(*Photo courtesy of Rossett School 2013*)

Rossett School's Valentine Message Service: Featuring hearts for two obvious reasons, Yr 9 students at Rossett School in Harrogate organised a Valentine's Day fundraiser. Heart-shaped sweets with messages attached were delivered around the school for £1 each. £85 was raised. Yr 9 chose BST as their charity after hearing about Philip Brown, 22/05/2005 — 29/09/2009.

Heart-shaped sweets for BST.
(*Photo courtesy of Rossett School 2013*)

(All photos in this section courtesy of BSTrust unless otherwise noted.)

Barth Syndrome Foundation of Canada
President's Report

By Lynn Elwood, President, Barth Syndrome Foundation of Canada

It is always heartwarming to look back over a year, and to see the successes and accomplishments that have been achieved, while making plans for the next. This year, the BSF of Canada celebrates its tenth year of existence, while we look forward to continuing key programs and exploring new ways to improve the lives of those affected by Barth syndrome and their families. We have seen a lot of growth and changes in the past years in the young men and families whom we serve. There has been more direct involvement by the affected individuals in the organization. We are grateful to them for their input which ensures that the programs we plan meet their needs.

During this year we have had deeper engagement with the International Barth Syndrome Foundation, as Lindsay Groff joined key board and executive meetings. There have been more joint projects and, of course, the conference. We are delighted that our Vice President, Cathy Ritter, has been named to the Barth Syndrome Foundation Board of Directors. Cathy brings great passion along with many skills to the BSF board and will continue in her current roles with the Canadian Board of Directors and Executive as well. We look forward to working with Cathy in her expanded roles in both areas.

Creating a yearly operating budget takes considerable thought and planning. With various fundraisers led by BSF of Canada and personal fundraisers, as well as careful expense control, we have achieved our overall financial goals for 2012. Here is our financial summary for the year.

Opening Balance	$91,309
Donations	24,795
Net Fundraising	22,448
Miscellaneous	2,124
Research Grant Funding	39,312
Program Expenses	21,593
Net Revenue	($11,538)
Closing Balance	**$79,711**

In the Science and Medicine program, we are delighted to once again be able to allocate funds to a research grant. To date, we have funded over $200,000 in Barth syndrome research, and this year we are participating in funding of a grant to Richard Epand, PhD, McMaster University, Hamilton, Ontario, Canada, entitled "**Relationship between membrane physical properties and the action of *tafazzin***". Raising awareness continues to be a priority for us, and we are making arrangements to participate in a grand rounds session in Winnipeg, Manitoba later on this year. We hope to connect and meet with families in the area while we are there.

We are very fortunate to have our own small army of dedicated volunteers. The list of jobs they do for us seems endless. It includes keeping the books, organizing and printing a Canadian newsletter, planning and doing prep work for our annual golf tournament, and holding personal fundraisers. A special group of volunteers are knitting a variety of items which will have our logo emblazoned on them. We are truly grateful to everyone for giving us their time and talents; we would not get very far without them. Over the past ten years our team approach has ensured the success of our organization and will continue to do so in the future. Thank you one and all.

BSFCa President Lynn leads us on the curling rink as well.

Nicki gives Robert falling advice.

Canadian Annual General Meeting
Business and Falling on Ice

By Robert, Affected Individual

Lynn presents award to volunteers Wayne and Dianne Bridger.

On 6 April, 2013, members of the Barth Syndrome Foundation of Canada (BSF Canada) descended on the Annandale Golf and Country Club for our Annual General Meeting (AGM), an afternoon of business and fun. The first part of the afternoon was spent reviewing the accomplishments of the BSF Canada in the past year, voting in board members, and hearing about the plans for this coming year. As always, attendees asked various questions during the meeting for any clarification and added their thoughts and comments. Near the end of the meeting, Wayne and Dianne Bridger, two of our dedicated volunteers, were presented with an award for their support and fundraisers for BSF Canada.

Once the meeting had adjourned, almost everyone moved to an adjacent curling hall where we tried, and mostly failed, to throw rocks into a bulls-eye on a sheet of ice. Thankfully we had secured a pro to give us some pointers, but even with Nikki's tutoring, balancing on one foot, with your weight partially on a heavy rock and partially on a broom, caused more than one person to fall. My personal curling career ended early on when I fell while stepping off the ice.

Adam cheers on Nikki, Travis & Lynn. 2013

Among the multitude of falls and rocks not going the distance, was Susan Hone's double takeout, securing victory for her rink.

Overall it was an enjoyable day, despite the pain from falling on the ice. I am looking forward to seeing everyone at this year's Golf Tournament in September.

Sheldon and Jacob plan strategy. 2013

Spotlight on the Families
Porter Ryan

By Ryan, Affected Individual

Porter Ryan. 2013

Ryan has successfully completed his first year of the two-year Culinary Management Diploma at Georgian College of Applied Arts and Technology. His full time studies included courses such as Food Theory, Kitchen Management, and of course many hours spent in the kitchen preparing, baking, grilling, sautéing and all the other skills required of a chef. He is currently honing his skills while working as a Porter in the kitchen at a local Golf Club. September will see him start his second year. His parents do wonder when he will start making supper for them at home however!

(Cont'd on pg. 21)

Spotlight on the Families

(Cont'd from pg. 20)

My Tattoo

By Joshua, Sibling of Affected Individual

I wanted to do something to remember my brother and also raise awareness for Barth syndrome. I decided a tattoo would serve both purposes. Jordan was my older brother who died in 1991, and Jared is my younger brother who is living with Barth syndrome.

Josh proudly displays his tattoo. [2013]

In Memory Of...

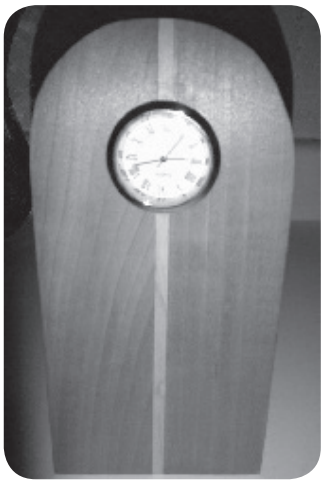
Moira Masterson

There have been some sad moments in the past few months, as two of our most staunch supporters, Moira Masterson and Elsie Morris, passed away. Moira, grandmother to Travis and friend to many, was an active volunteer for many years, selling poinsettias, raising awareness, and joining Canadian and international events. Elsie, great-grandmother to Adam, was always there in the background, and contributed to every fundraiser that was brought to her attention. As a final wish, both Moira and Elsie named the BSF of Canada as their charity of choice, and both were honoured with memorial donations. To the families of these two ladies, we send our heart-felt condolences. Their support and dynamism will be missed by all.

Elsie Morris

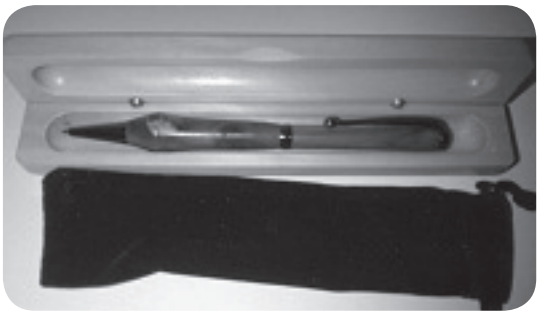
"Friendraising" Fundraising by BSFCa

Toward the end of last year, we were the lucky recipients of three fundraisers held by our members. Once again, Cathy Ritter sold poinsettias. She has held this fundraiser yearly, and people look forward to buying these beautiful plants which bring warmth and colour to their homes during the Christmas season. Audrey Hintze, who has made many contributions to our organization, held a seasonal gathering of friends in December and collected donations. Lynn Elwood ran a new fundraiser also in December. Woodworking prizes were made by Les Morris, Lois Galbraith, and Barth affected individual, Adam. Lynn and friends sold raffle tickets raising an amazing $1,000. Thank you to everyone for their contributions. It was a great way to end the year, not only by giving our finances a boost, but also by raising awareness.



Cherry Desk Clock won by Ernie Jones.

Nesting Pine Tables won by Reg Hamilton.



Juniper Pen won by Stephan Ludlow.

(All photos in this section courtesy of BSF Canada [2013])

Association Barth France 2012 Research Funding

By Florence Mannes, Chair, Association Barth France

In 2012, Barth France financially supported two different research programs, one French and one American:

FRENCH PROJECT: Natural History of Barth syndrome: A National Cohort Study of 22 Patients

This study includes 22 Barth cases that have been identified over time in France, 10 of whom were alive at the date of publication.

Abstract:
"We identified 16 BTHS pedigrees that included 22 patients. *TAZ* mutations were observed in 15 pedigrees. The estimated incidence of BTHS was 1.5 cases per million births (95%CI: 0.2–2.3). The median age at presentation was 3.1 weeks (range, 0–1.4 years), and the median age at last follow-up was 4.75 years (range, 3–15 years). Eleven patients died at a median age of 5.1 months; 9 deaths were related to cardiomyopathy and 2 to sepsis. The 5-year survival rate was 51%, and no deaths were observed in patients ≥3 years. Fourteen patients presented with cardiomyopathy, and cardiomyopathy was documented in 20 during follow-up. Left ventricular systolic function was very poor during the first year of life and tended to normalize over time. Nineteen patients had neutropenia. Metabolic investigations revealed inconstant moderate 3-methylglutaconic aciduria and plasma arginine levels that were reduced or in the low-normal range. Survival correlated with two prognostic factors: severe neutropenia at diagnosis (<0.5 × 109/L) and birth year. Specifically, the survival rate was 70% for patients born after 2000 and 20% for those born before 2000.

Conclusions: This survey found that BTHS outcome was affected by cardiac events and by a risk of infection that was related to neutropenia. Modern management of heart failure and prevention of infection in infancy may improve the survival of patients with BTHS without the need for heart transplantation."

U.S. PROJECT: Cardiolipin deficiency leads to defects in the TCA cycle

Building on the tricarboxylic acid (TCA) cycle dysfunction hypothesis of Barth syndrome as put forth by Dr. Richard Kelley, Dr. Miriam Greenberg will examine the TCA cycle in yeast mutants that are compromised in their cardiolipin expression, which includes the *tafazzin* deletion strain. Dr. Greenberg has provided unpublished data to show that mitochondrial dysfunction caused by cardiolipin alterations involves and impacts the proper functioning of the TCA cycle — the pivotal metabolic system of mitochondria-containing cells. Dr. Greenberg will measure: metabolite levels, enzyme activities, mitochondrial retrograde pathway gene expression (the pathway of proteins and metabolites whereby the mitochondria communicates with the nucleus of the cell to alter metabolism, i.e. *tafazzin*, TCA cycle enzymes, TCA cycle intermediates, etc.), beta-oxidation pathway (metabolism of fats), and the glyoxylate cycle (a short-circuit of the TCA cycle not found in animals). In addition, the supplementation of oleic acid to the growth media rescues yeast cardiolipin mutant strains, and uncovering the basis for this effect will be investigated. Dr. Greenberg hopes that by identifying TCA cycle abnormalities we will better understand how anapleurtoic/nutritional supplements (like arginine) may be beneficial for the treatment of Barth syndrome.

June a Busy Month for Barth France

June 14: Gospel Concert Colors

On June 14th, the members of the Gospel Colors choir gave a concert at the Church of Saint Honoré d'Eylau's for the benefit of Barth France and the great pleasure of the audience. Their energy and music naturally engaged the audience and there were lots of smiling faces at the end of the show!

Created in 2004, Gospel Colors is a very dynamic and non-religious choir, whose members are either amateurs or volunteers, united by their common passion, Gospel. The choir gives concerts in Paris for charity or humanitarian causes. Each member of the audience is free to be involved.

(Cont'd on page 24)

Association Barth France

By Florence Mannes, Chair, Association Barth France

Barth France a financé, en 2012, deux programmes de recherches, l'un français, l'autre américain.

ETUDE FRANÇAISE: Histoire naturelle de la maladie de Barth à partir de l'étude de la cohorte des patients français
Cette étude a permis d'identifier, en France, 22 cas atteints du Syndrome de Barth, dont 10 sont en vie à la date de la publication de ce journal; l'analyse de leur histoire médicale a donné lieu à une publication dans le Orphanet Journal of Rare Diseases.

Ci-après une synthèse de cet article:
«Nous avons identifié 16 pédigrées du Syndrome de Barth, qui englobent 22 patients. La mutation du gène *TAZ* a été observée dans 15 de ces pédigrées. L'incidence estimée du Syndrome de Barth est de 1,5 cas par million de naissance. L'âge médian d'apparition des premiers symptômes se situe à 3,1 semaine (pour des résultats allant de la naissance à 1,4 an), et l'âge médian des patients lors de leur dernier suivi était de 4,75 ans (l'âge au dernier suivi s'étalant de 3 à 15 ans). Onze patients sont morts à un âge moyen de 5,1 mois; 9 décès sont liés à une cardiomyopathie, 2 à des chocs septiques. Le taux de survie à 5 ans est de 51%, et aucun décès n'a été observé chez des patients âgés de plus de 3 ans. Quatorze patients présentaient une cardiomyopathie (…). La fonction systolique du ventricule gauche était généralement très mauvaise pendant la première année de vie, et tendait à se normaliser une fois passée cette période critique. Dix-neuf patients étaient atteints de neutropénie. Les investigations métaboliques ont permis de révéler une acidurie methylglutaconique modérée et non constante, ainsi que des niveaux d'arginine dans le plasma bas, ou dans la norme basse. La survie est corrélée avec deux éléments: l'importance de la neutropénie lors du diagnostic, et l'année de naissance. En particulier, le taux de survie à 5 ans est de 70% pour les patients nés après 2000, alors qu'il était de 20%pour ceux nés avant 2000.

En conclusion, cette étude a permis de montrer que les conséquences du Syndrome de Barth étaient liées à la santé cardiaque des patients ainsi qu'aux risques d'infection, liés à la neutropénie. Une gestion moderne de l'insuffisance cardiaque ainsi que la prévention des infections durant la petite enfance peuvent améliorer le taux de survie des enfants atteints du Syndrome de Barth, sans devoir avoir recours à une transplantation cardiaque de façon systématique.»

PROJET AMERICAIN: Impacts de la carence en cardiolipine dans le dysfonctionnement du cycle de Krebs
Cette étude, qui fait partie de la dizaine de programmes de recherche sélectionnés par le comité scientifique de la Barth Syndrome Foundation, et conduite par le Professeur Miriam Greenberg, PhD à l'Université de Detroit, part de l'hypothèse, mise en avant par le Dr. Kelley, que le cycle de Krebs (série de réactions biochimiques dont la finalité est de produire des intermédiaires énergétiques) connait, chez les patients atteints du Syndrome de Barth, un certain nombre de dysfonctionnement. Dans cette étude, le Pr. Greenberg va étudier le cycle de Krebs de levures génétiquement modifiées pour avoir un défaut en cardiolipine. Par cette étude, le Dr. Greenberg espère, en isolant les anomalies du cycle de Krebs chez les patients atteints du Syndrome de Barth, avoir une meilleure compréhension de l'efficacité des compléments alimentaires (arginine) dans le traitement du Syndrome de Barth.

C'est le 7ème programme de recherche du Dr. Greenberg sur le Syndrome de Barth; le Dr. Greenberg connaît très précisément les mécanismes propres à cette maladie. Cette étude a donné lieu à de nombreuses présentations ainsi qu'à la publication de trois articles médicaux ("Lipidomics of intact mitochondria", "Loss of cardiolipin leads to perturbation of mitochondrial and cellular iron homeostasis", "Cardiolipin-mediated cellular signaling".

Un mois de juin bien rempli pour Barth France

14 juin : Concert Gospel Colors

Le 14 juin dernier, les chanteurs de la chorale Gospel Colors ont fait un concert en l'Eglise saint Honoré D'Eylau au profit de l'association Barth France pour le plus grand bonheur des spectateurs présents. Au programme, une énergie musicale qui se communique naturellement à l'assemblée et beaucoup de sourires à la sortie du spectacle!

Créée en 2004, Gospel Colors est une chorale très dynamique, laïque, formée de chanteurs amateurs et bénévoles réunis autour d'une passion commune, le Gospel. Elle se produit en concert à Paris, en faveur d'associations caritatives ou humanitaires, chaque spectateur étant invité à donner une participation libre.

(Suite à la page 25)

June a Busy One for Barth France

(Cont'd from page 22)

June 16: Course des Héros

For the third year Barth France participated in "la course des héros." Only members who have collected at least 200 euros (sometimes they collect a lot more) can participate in this run for the association they chose to represent, and every year it is a big event for Barth France.

There were 12 runners who chose to join us in this 6 kilometer run this year at the Parc de Saint Cloud and to represent Barth France with a special thought for Pierre. Congratulations to Stéphy, Stéphanie, Florence, Isabelle, Martine, Julie, Valerie, Romain, Antoine, Hélène, Emilie, and Yves!

June 18: Exhibition of works of art

It is often said that it is all about who you meet... This happens to be true, especially when you are involved in a charity… Ana Perez Grassano is a friend of a friend.... A mother whom I met fetching my kids to school. She's an architect, a business woman who lives in the fast lane....but Ana is not just that, she's also a big hearted artist. When she organized her first exhibition at her home with four other artists, Maria Burghetto, Rafael Gimenez, Isabelle Castagné and Christine Beroff, she convinced them to donate a portion of the benefits to Barth France… After her big success, she decided to repeat the experience with a friend of ours, Sophie Dréan, who we didn't know was also a talented painter.

During this second exhibition, they sold almost all of their paintings, and gave more than 1000 euros to Barth France. Because Ana is a woman of her word and a very involved person, she organized another exhibition on June 17th and 18th, at the Argentinian Ambassy in Paris, with famous artists Richard Orlinski, Guillaume Saint Michel, and Paz Alvarez Mendendez.

In Loving Memory of Pierre

See page 25 for the French translation of this article.

Pierre Bruel

"Pierre passed away on May 23rd from a throat infection." It's with these simple words and with a great sense of dignity that Christine and Benoit, announced yesterday, the death of Pierre. He would have been 10 years old next September.

Pierre's death reminds us all that Barth syndrome is a serious disease from which one can die at only nine years old because of a weak immune system.Pierre had Barth syndrome and was diagnosed very early. He had a heart transplant when he was still a baby and would have celebrated his 8-year transplant anniversary. Pierre had a normal life despite his medication. Pierre was going to school, eating at the canteen, and also cycling.

On May the 23rd, Pierre had a fever. The doctor diagnosed a throat infection and sent him to the hospital. Pierre passed away a few hours after that. In only one day, an entire family's life collapsed… It has been so sudden, so senseless, so hard… how can anyone die from a throat infection, and so young…How can someone possibly survive a heart transplant at two and die from a throat infection at ten? Why are bacteria still faster than the medicine?

The epidemic study financed by Barth France last year showed that: in France, Barth syndrome affected 22 children, eleven were alive, now only 10 remained… Behind those numbers hides a striking reality that gives even more sense and purpose to Association Barth France's work.

Pierre, our association misses you. We think of your parents, your sister, Manon, and your brother, Quentin, for whom the loss is huge. Pierre, you give us the strength to keep on fighting! We will not give up!

Un mois de juin bien rempli pour Barth France

(Suite de la page 23)

16 juin: Course des Héros

Pour la troisième année consécutive, Barth France était présent à la course des héros. Cette course, qui ne regroupe que des personnes ayant réussi à collecter au moins 200 euros (et parfois beaucoup plus) pour l association de leur choix, est un événement marquant de l'année pour Barth France.

Ils étaient donc 12, cette année, au Parc de Saint Cloud, à s'être engagés à nous suivre, sur les 6 km de la course, sous les couleurs de Barth France, avec notamment une pensée toute particulière pour Pierre. Bravo à Stéphy, Stéphanie, Florence, Isabelle, Martine, Julie, Valerie, Romain, Antoine, Hélène, Emilie et Yves!

18 juin: Exposition d'œuvres d'art

On dit que tout est souvent affaire de rencontres.... Et cette phrase se vérifie particulièrement quand on s occupe d une association .... Ana Perez Grassano est une amie d'amis....une maman croisée à la sortie de l'école, une architecte, femme d'affaires vivant à 100 à l'heure....mais Ana n'est pas que cela, c'est aussi une artiste au cœur grand comme ça....lorsqu'elle décide d'organiser sa toute première exposition, chez elle, avec 4 autres artistes (Maria Burghetto, Rafael Gimenez, Isabelle Castagné et Christine Beroff), elle les convainc de reverser une partie de ventes des tableaux à Barthfrance .... Forte de ce succès, elle renouvelle l'expérience, avec une autre amie (Sophie Dréan), amie commune qui nous avait jusque la caché ses talents de peintre.

Lors de cette deuxième exposition, elles ont toutes deux vendu la quasi intégralité de leurs tableau, et on reverse à Barthfrance plus de 1000 euros. Et, parce qu'Ana est quelqu'un de parole et de conviction, elle organise le 17 et 18 juin, une nouvelle exposition, d'une toute autre ampleur, à l'ambassade d'Argentine, avec des artistes renommés (Richard Orlinski, Guillaume Saint Michel et Paz Alvarez Mendendez). Si cette exposition a permis de collecter des dons, cela a également été l'occasion de faire connaître le Syndrome de Barth à un large public.

En mémoire de Pierre

Voir les page 24 pour la traduction anglaise de cet article.

Pierre Bruel

«Pierre nous a quitté le 23 mai des suites d'une angine,» c'est par ces mots simples et avec beaucoup de dignité que Christine et Benoit, ses parents, nous ont annoncé le décès de Pierre. Pierre aurait eu 10 ans en septembre.

Le décès de Pierre nous rappelle à tous ce qu'est le syndrome de Barth, une maladie grave dont on peut mourir à 9 ans par manque de défense immunitaire. Pierre était atteint du syndrome de Barth, diagnostiqué très tôt. Il avait subi une transplantation cardiaque dès le plus jeune âge et s'apprêtait à fêter les 8 ans de sa greffe. Pierre menait une vie tout à fait normale malgré ses traitements. Pierre allait à l'école, Pierre mangeait à la cantine, Pierre faisait du vélo.

Le 23 mai, Pierre a eu de la fièvre. Le médecin a diagnostiqué une angine et a envoyé Pierre à l'hôpital. Pierre est parti en quelques heures des suites de l'angine. En une journée, la vie d'une famille s'écroule….C'est tellement soudain, tellement absurde, tellement dur....comment peut-on mourir d'une angine, quand on n'a pas encore 10 ans...comment peut-on survire à une transplantation cardiaque avant l'âge de 2 ans, et mourir d'une angine? Pourquoi les bactéries sont-elles encore plus rapides que les médicaments…

L'étude épidémiologique financée par Barth France l'an dernier annonçait une statistique: en France, le syndrome de Barth c'est 22 cas recensés dont 11 enfants en vie, c'est désormais 10 enfants…. Derrière ces chiffres se cache une terrible réalité qui donne encore d'avantage de sens et de motivation à l'action de l'association Barth France.

Pierre, tu manques à notre association. Nous pensons très forts à tes parents, à ta sœur Manon ainsi qu'à ton frère Quentin, pour qui le vide doit être immense. Pierre, tu nous donnes le courage de nous battre! Nous n'abandonnerons pas.

Barth families, have you moved lately? Please help us keep your information current.

In the past, the Post Office notified us of address changes. However, with so few actual mail pieces being sent during the year, we will not know you have moved unless you tell us. If your telephone number and/or email address has changed, please let us know. If we do not have your email address, please go online to add it to your contact information.

Barth families, if you think any information on your family might be incorrect, please be sure to update us.

Visit BSF's website and complete the **Contact Information** form that can be found under Families >> Update Contact Information (http://www.barthsyndrome.org/english/View.asp?x=1568). Thanks in advance for helping us "keep house."

Levi (age 1)

Wyatt (age 5)

(Photos courtesy of BSF ~ 2013)

Donations Made Easier

Donate by check: Make check payable to **Barth Syndrome Foundation, PO Box 582, Gretna, NE 68028**

Donate online: You can donate to BSF or any of the international affiliates by going to our website, www.barthsyndrome.org, and clicking on the "Support BSF" link on our home page, or through Network for Good (https://www.networkforgood.org/donation/ExpressDonation.aspx?ORGID2=22-3755704) or through Paypal (https://www.paypal.com/cgi-bin/webscr?cmd=_s-xclick&hosted_button_id=8XRHKG52LB7L4).

Donate through Causes on Facebook: Join us on our online social network (http://www.causes.com/causes/46297-the-barth-syndrome-foundation?q=barth+syndrome+foundation&rank=0&utm_campaign=search).

Employer Matching Gift Programs: Many donors are now taking advantage of a "Matching Gift Program" offered by their employer. The employer matches the funds donated by the employee to a charity and provides a convenient method for the employee to donate to a charity of his/her choice.

Planned Giving: One of the best ways to support our continued efforts is to remember BSF (or its affiliates) in your estate planning. Talk to your lawyer or estate planning professional about including BSF (or its affiliates) in your will.

BBB ACCREDITED CHARITY
bbb.org/charity

Power of Kindness

Donor categories are based upon the past 18 months of cumulative giving

Barth Syndrome Foundation

PAULA & WOODY VARNER FUND

Angels ($1,000 - $4,999)
Dillon Foundation
Henricks, Dr. Bruce & Peggy
Sonderegger, Ted & Mary Ann
Wilkins, Sue & Dr. Mike

General Contributions ($50 - $999)
Acklie Charitable Foundaiton
Allman, Tom & Jane
Allman, Peter & Maureen
Basler, Dr. Rod & Debbie
Bettenhausen, Bob & Joanne
Beynon, Dave & Liz
Bingham, Dr. Dave & Kathy
Buckley, Les & Nancy
Burmeister, Blaine & Abby
Burmeister, Charles & Marita
Burmeister, Jon & Jackie
Campbell, Bob & Candy
Capek, Dr. Dale & Clay
Carveth, Dr. Steve & Beth
Cent, Norb & Bev
Cheatham, Dr. John
Cheatham, Linda
Cimino, Dr. Mike & Patty
Desmond, Jerry & Sally
Dittman, Alice
Farrar, Doug & Shawn
Firestone, Dave & Jane
Ganz, Doug & Pam
Gelber, Dr. Ben & Elaine
Haessler, John & Nancy
Harford, Kevin & Sonia
Hedgecock, Norm & Debbie
Henricks, Dr. Bruce & Peggy
Jensen, Tom & Marilyn
Keating, Con & Barbara
Kiechel, Dr. Fred & Vivian
Linder, Dr. Max & Pat
Massengale, Dr. Martin & Ruth
McNair, G. L.
Minnick, Gates & Daisy
Nelson, Scott & Teri
Norris, Dr. Mike & Mary
Olson, Loy & Julie
Osborne, Dr. Tom & Nancy
Otte, Robert & Carolyn
Pittenger, Jim & Julie
Rawley, Ann
Roskens, Dr. Ronald & Lois
Schorr, Paul & June
Seaman, Andrew & Robyn Steely
Sonderegger, Kurt & Sue
Stohs, Dr. Gene & Kristen
Stuart, James & Susan
Stuart, Jr., James
Stuckey, Dennis & Nancy
Swanson, Mary
Tegt, Dr. Tom & Barb
Thompson, Elwood & Carol
Varner, Tom & Beth
Varner, Judy
Walker, Kennedy & Dianne
Wiederspan, James & Ann
Wiederspan, John
Wilkins, Dr. Jerry
Wilkins, Joanne
Wilkins, John
Wilkins, Dr. Lee & Kristi
Wilkins, Muriel

SCIENCE & MEDICINE FUND

Stars ($5,000+)
Allen, Tiffini
McCurdy, Steve & Kate
Sernel, Marc & Tracy

Angels ($1,000 - $4,999)
Clark, Robert (Seeds Of Hope)
Clark, Bob & Randy Galbreth
Kirkland and Ellis Foundation
Kugelmann, Peter & Karen
Kugelmann, Steve & Jan
Winston, Laura

General Contributions ($50 - $999)
Allen, John & Karen
Baron, John & Christine
Barrett, James & Nancy
Caille, Jan & Karen
Campbell, Tonja
Clark, Jeffrey
Davis, Samuel & Susan
Dollard, John & Janet
Foster, Christine
Gilbert, Gregory
Harrigan, Timothy & Lori
Keller, Allan
Lloyd-Jones, Kimball & Mary
MacDonald, Grant
Mansfield, Anne
Nishi, Michael
Nurkowski, John & Tina
Olson, Richard & Sharon
Roberts, John & Betty
Robertson, Tracy
Robinson, Joe & Connie Sue
Ryan, James & Miriam
Stahly, Craig (Priority One Construction)
Watson, Theda
Wayne, Alexander
Wiley, Troy & Gracie
Yancy, Eric

GENERAL FUND

Stars ($5,000+)
American Express PAC Match
Cusack, Tom & Carrie
The Frill Foundation
Grubb & Ellis
The Lebensfeld Foundation
Lummis, Marilyn
Lummis, William & Dossy
The Malkin Fund
Malkin, Scott & Laura
McKown, Christopher & Abby Johnson
Miller, Kirt & Martha
Pierson, Dr. Richard & Allene
Allene & Paul Russell Fund
Russell, Trip & Sara
Sernel, Gordon & Sharon
Earl and Brenda Shapiro Foundation
Stoll, Ned & Cindy

Angels ($1,000 - $4,999)
Anonymous
Arquit, Kevin
Association Barth France
BSF of Canada
Bailey, Clark & Patricia
Baker, Timothy
Barad, Seth & Amy
Buddemeyer, Randy
Buly, Dr. Robert & Lynne
Condon, Owen
The Martha Davis & Richard Sayrs Family Fund
Dell Giving
Despins, Suzanne
Dooley, Adam
Epstein, Paul & Leah
Evoy, Larry
Forsyth, John
Susan & David Fowler Charitable Gift Foundation
Galbraith, Lois
Geary, Ann
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(Cont'd on page 28)

Rachel and Jack (age 2) 2012

Ned and Milosh (age 3) 2012

Tiffini and Henry (age 4) 2012

Chris and Christopher (age 5) 2012

(Photos courtesy of Amanda Clark ~ BSF 2012 Conference)

Power of Kindness

" Our son is affected by this disease and when he was diagnosed over 10 years ago, the Barth Syndrome Foundation dramatically changed his path of treatment and has been a life line for us. The medical advisory staff is incredible! The people involved and the families affected are extraordinary! The foundation provides instant access of help from other families as well as experts in the disease and can truly save lives by just one phone call, text, or email!"
~ Affected Family, Great NonProfit Review, 2013

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Power of Kindness

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Hindriks, Alberdine
Hinton, James
HMRC Staff, Cardiff
Holdsworth, Daphne
Holmes, Pam & Rowland
Holmes, Samantha
Homewood, Alison
Hope, Chris & Robert
Horwood, Jane, Catfish Web Design
Howard, Chris
Howe, Malcolm
Howell, H
HSBC, Bristol
Inder's Kitchen
Jenkins, Don
Jones, Cynthia
Jones, David
Jones, Lucy
Jones, Patricia
Jones, Patrick
King, Mrs J, Misses P & S
Kings' School, Winchester
Koehler, Richard
Koh, Amelia
KSC Phoenix, Jay's Football Team
Lallemand Family
Lange, Maike & Alan Brown
Latham, Daisy
Legal & General, Cardiff
Lendrum, Caroline
Lester, Jacqui
Leveson, Patricia
Longhurst, Ally
Loo, Jo & Albert van
Majumdar, Dr Arnie
Mannes, Florence & Philippe
Manor Booze
Manton, Annick & Rob
Manton, Greg & Kerry-Ann
Manton, Joan
Manydown Farm
Marley, Valerie
Martin, Dr Robin
Martin, Mrs L
May, Derek
McGregor, D
Merce , Mrs K
Michener, Kathryn
Moore, Nigel & Lorna
Morby, Hannah
Mumford-Rudd, Amanda & Allison
Murphy, Helen
Murray Craig
Nelson, Laidley
Newbury-Ecob, Dr. Ruth
Nicholas, Hannah
Nicol, Charlotte
Norgate, Mrs Christine @ Kings' School
Nyack, Adrian
Oakley Tennis Club
Olden, Karen
Olden, Lee
Oram, Richard & Heather
Overton Methodist Church
Overton Tennis Quiz Team
Pants, Candice
Parkhurst, Yolande
Payne, Jason
Pettigrew, Veronica
Phillips, Alan & Bridget
Piette Feck, Astrid & Arthur
Play Centre Staff, Bristol Royal Hospital for Children
Polar Bear Windows of Bristol
Porter, Bev
Prest, Simon
Ratzker. Nicole
Rawlinson-Millichap, Dominic & Peter
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Reddin, Mick & Judi
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Riddiford, Debbie
Riseborough , Ms K A
Riseborough, Mr & Mrs L
Riseborough, Leslie R.
Riseborough, Mrs Thelma
Roberts-Collins, Cara
Rogers, Rev Dr Sarah
Rook, Joyce, Newton St Cyres Meat Bingo
Roseberry Sports and Community College
Rossett School, Yr 9
Royal Gwent Hospital, Staff
Ruzicka, Helen
Salmon, Gwen
Salmon, Lawrence
Salmon, Robertina
Salon at No 5
Schachat, Rally
Schlapak, Gregor & Sonja
Serra-Palmer, Mrs M
Setton, Amanda
Sharpe, Charlotte, Nick & Family
Shaw, Eddie
Shepreth Wildlife Park
Smith, Brenda & Wilf
South Wales Mountaineering Club
St Catherine's Church, Caerphilly
St Dunstan's Church Community Café, Ashurst Wood
Stagg, Lizzie & Kevin
Steele, Kenny & Alexia Drakakis
Steward, Dr Colin & Christine
Stinchcombe, Dennis, Louis & Michelle
Stobart-Hook, Barry & Nicolette
Stobart-Hook, Guy & Liz
Stone, Dr Janet
Swennen Family
Symonds, Alistair
Taylor, Sarah
Temple, Ailsa
Thomson, Nigel
Thorne, Antony
Thorpe, Astrid
Thorpe, Jeremy
Thorpe, Nigel
Thorpe, Thomas
Thyne, Hunter
Timerick, Jo
Todd, Ian
Treasury Wine Estates, Australia
Tsai-Goodman, Dr Beverley
Upfold, Rachel
Verdon, Shelene
Wald, Amy
Ward, Anne & Barry
Ware, Jenny
Warner, Rosemary
West Family
West, Claire
Westgate Primary School
White, George
White, Mark
Whithorn, Sarah
Williams, Mary
Williams, Norman
Winn, Melese
Wites, Wojciech & Agnieszka
Woodward, Tracy & Ian
Woolley, Scott & Julie
Wright, Jess

Jack (age 7) 2012

Alex (age 12) 2013

Ieuan (age 16) 2013

Power of Kindness

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Hope, Harry & Helen
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Hope, Robert
Hope, Terry & Ruth
How, Jimmy
Howard, Sharon
Hu, Ian
Hubble , Larry
Hucaluk, Adrienne
Hughes, Ron & Cathy
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Husar, Vandra
Ian, Hu
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Jardine, Ward
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Jones, Marilyn
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Kostal, Jerry
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Malick, Mel & Suzanna
Mallais, Mary & Raymond
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Marchesin, Elvy
Marcucci, Gabe
Maritz,
Markham, Wanda
Martin, Dave
Martin, Margaret
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McCrodden, Jack
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McDowell, Joan & Bill ~ J & B Customs Brokers Ltd.
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Moore, Nigel
Moore, Walt
Morris, Elsie
Morris, Erik
Morris, Ian ~ JonesDeslauriers
Morris, Lois Galbraith & Les
Moss, Don
Muller, Dea
Murphy, Tony
Musgrave, Bruce
Nagel, Siegfried & Erna
Nagel, Thomas
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Robinson, Val
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Schertzl, Dawn
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Schuller, Dana
Schuller, Dennis ~ Aviall Canada Ltd.
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Segal, Joseph
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Simms, Marie
Simsons, Joyce
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Storey, Dick & Janis
Storey, Jan
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Teva
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Thomas, Val
Thompson, Diane
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Tyers, Robert ~ Wellington Consulting Inc.
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Weaver, Joan
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Wegman, Gelja
Weller, Sharon
White, Susie
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Wilks, Dennis
Wilks, Helen
Willis, Dave & Penny
Windrim, Brenda
Wiwcharyk, Lubow
Wood, Gale
Woodcock, Sharon & Roy
Worsley, Dorothy
Wu, Jin
Wuthrich, Grace
Young, Joan
Young, Ron & Lenora
Zavitz, Peter

Adam (age 23) 2013

Sheldon (age 12) 2013

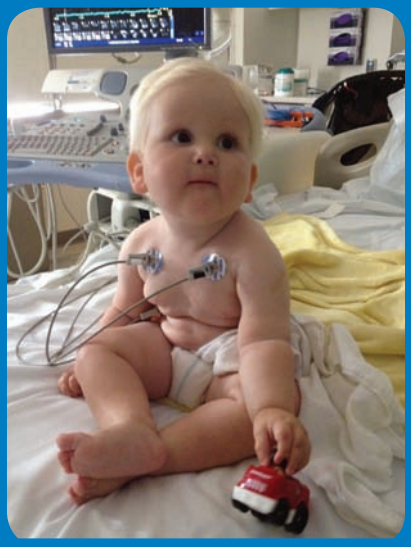
Robert (age 27) 2013

Barth Syndrome Foundation
PO Box 618
Larchmont, NY 10538
Phone: 850-273-6947
Facsimile: 518-213-4061
Email: bsfinfo@barthsyndrome.org
Website: www.barthsyndrome.org

Barth syndrome (BTHS; OMIM #302060)

A rare, serious, genetic disorder primarily affecting males. It is found across different ethnicities and is caused by a mutation in the *tafazzin* gene (*TAZ*, also called G4.5), resulting in a complex inborn error of metabolism.

Though not always present, cardinal characteristics of this multi-system disorder often include combinations and varying degrees of:



Connor (age 2) [2013]

- **Cardiomyopathy** *(usually dilated with variable myocardial hypertrophy sometimes with left ventricular noncompaction and/or endocardial fibroelastosis)*
- **Neutropenia** *(chronic, cyclic, or intermittent)*
- **Underdeveloped skeletal musculature and muscle weakness**
- **Growth delay** *(growth pattern similar to but often more severe than constitutional growth delay)*
- **Exercise intolerance**
- **3-methylglutaconic aciduria** *(typically a 5- to 20-fold increase)*
- **Cardiolipin abnormalities**

For more information, please visit Barth Syndrome Foundation's website:
www.barthsyndrome.org