



Barth Syndrome
Foundation

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

Enthusiasm for Barth Syndrome 2014 Conference

By Hilary Vernon, MD, PhD, McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University; Kennedy Krieger Institute, Baltimore, MD

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Dr. Hilary Vernon

I am looking forward with great enthusiasm to the upcoming Barth Syndrome Foundation International Scientific, Medical and Family Conference in June 2014.

I became involved with the Barth Syndrome Foundation (BSF) two and a half years ago, and attended my first BSF International Scientific, Medical and Family Conference in June 2012. At that meeting, my research team from the Kennedy Krieger Institute conducted a study looking for biochemical and other laboratory factors important in Barth syndrome.

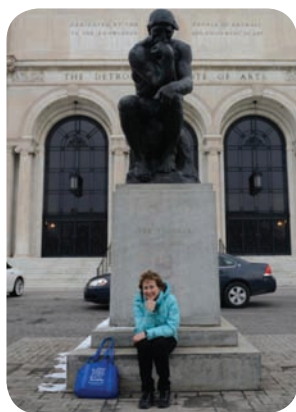
Due in large part to the enthusiastic participation of the members of the Barth syndrome community, we were able to gather substantial evidence for a unique biochemical profile in Barth syndrome. This work has just been published in the journal *Molecular Genetics and Metabolism*, which

is widely read by the biochemical genetics community. We will build upon this work at the upcoming meeting, by gathering further biochemical data and clinical data.

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BSF Seed Funding Leads to NIH Grants

By Miriam L. Greenberg, PhD, Biological Sciences, Wayne State University, Detroit, MI



Dr. Miriam Greenberg

Scientific research elucidates fundamental knowledge that can be utilized in applied technologies to improve our health and quality of life. Few would disagree that it is in our best interests to support medical research and develop cures for devastating diseases. Nevertheless, federal funding for research has been steadily declining, and the National Heart, Lung, and Blood Institute of the National Institutes of Health (NIH) is currently funding only 10% of submitted applications. Many outstanding and creative proposals are not funded, and scientists are forced to spend inordinate amounts of time trying to obtain grants. During this very bleak funding climate, support from non-federal sources is often crucial for scientists to test new hypotheses and develop new paradigms that may lead to important breakthroughs in medical research.

Support from the Barth Syndrome Foundation (BSF) has enabled me to carry out Barth syndrome-focused research that has led to subsequent NIH funding. My first BSF grant from the 2002 grant cycle ("**TAZ1 Gene**

Function in Yeast: A Molecular Model for Barth Syndrome") supported the development of the yeast model for Barth syndrome. This laid the groundwork for all of our subsequent studies of *tafazzin*-deficient yeast cells and helped us to obtain the preliminary data leading to an NIH R21 grant (2008-2010) entitled "**Synthetic Lethal Interactions in Barth Syndrome.**"

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Growth and Progress Because of You

By Lindsay Groff, Executive Director, Barth Syndrome Foundation



Lindsay Groff

Progress and growth — this is what will come to mind when you read through the incredible articles in this edition of the journal. I want you to take great joy in knowing the essential role you play in all of this success. Progress is evident in our research grant program. In addition to the worthy grants funded in this cycle, Dr. Miriam Greenberg explains how seed funding, given from BSF over 10 years ago, has led directly to a much larger grant from the National Institutes of Health. Our “small” research grants make such a difference in this tight funding climate by launching and giving credibility to research that might never get funded at all.

It's incredible to think that our seed funding 10 years ago is now helping Dr. Greenberg take her research to a whole new level...a level that may lead to new discoveries in helping those affected by Barth syndrome.

Growth is abundant in planning our 2014 Conference in Clearwater, Florida. We anticipate record-breaking attendance this year with 50 affected individuals registered! Imagine a boy or young man who may be the only person in his entire state or country living with Barth syndrome coming together with 49 “brothers” in one place. Bringing the families together with the world-renowned experts in Barth syndrome is nothing short of amazing.

For clinicians and scientists, this Conference also serves to accelerate progress as the latest research is presented and new ideas are discussed. New insights are gained and collaborations are fostered, both of which catalyze scientific and medical advances.

Many first-time attendees describe the Conference as life-changing. For the families, the information gained from the doctors, researchers, and other parents helps provide answers for this ultra-rare disorder. The clinicians and researchers gain a new appreciation for our cause as they interact with the boys and young men in a social setting.

Funding all of this important progress and growth takes hard work. This year we set an ambitious goal to raise \$1,000,000. Yes, one million dollars! That represents almost a 20% increase over last year. As we have grown, the board and I have agreed we needed to hire someone to work full-time with fundraising. In March, we hired Sandra Stevens to help coordinate and execute many of our fundraising programs. She has already made an impact in just her short time with us. To learn more about Sandra and the new efforts underway see page 14.

Why is our goal so aggressive in 2014? Because, right now, there are some amazing opportunities for funding scientific and medical research on Barth syndrome...opportunities that are unprecedented. Our hope is that these opportunities will lead to breakthroughs... breakthroughs that will make the lives of our boys and young men easier and better. This is what it's all about. That is exactly how we will continue with the progress, growth, and hope.

However, in order to meet our goal this year we are going to need your continued support. Without you, there is no Barth Syndrome Foundation. Without you, the research doesn't happen. Without you, there would be little hope. Don't be surprised if you hear from me or Sandra a little more often asking for your support. More importantly, know that if you ever have a question or concern, you can give me or Sandra a call. We would love to hear from you.

Thank you again for all you do for the boys and young men with Barth syndrome. One day there will be a cure — everything you are doing is helping to make that day come sooner.

Adjusting To a New Normal

By Marc Sernel, Chairman, Barth Syndrome Foundation

Out of my comfort zone... That is where I find myself when I sit down to write this personal piece as Chairman of the Barth Syndrome Foundation (BSF) Board. I actually like to write and do a lot of it in my "real job" as a patent and trial lawyer. But writing to persuade a judge or client of some legal argument is very different from writing to the diverse group of people across the globe that forms our Barth Syndrome Family, so please bear with me.

Adjusting to a "new normal" is something those of us touched by Barth syndrome have more experience with than most. Although sometimes we can't help but lament the fact that the "old normal" may no longer be an option, we all try to find and cherish our own new normal and soldier on. Sometimes that means finding some semblance of a normal routine during a long hospital stay or coping after the tragic loss of a loved one. We strive to embrace what we do have and hopefully find even greater fulfillment in making the best of the hand we have been dealt. I have great respect for, and draw inspiration from, our Barth boys and men because they focus on what they *can* do, not on their limitations or what they can't do.

Like our Barth guys, this organization is continually adjusting to an ever-evolving "normal." And this is a good thing. The organization has come a long way since its creation in 2000. I dare say that no one on the founding Board could quite have imagined then what BSF has become today. That first governing group set a goal, incredibly ambitious at the time, to raise \$750,000 over five years. They subsequently blew past that and set the bar even higher. In 2013 alone, we raised over \$800,000, and we are going to work really hard to break through the \$1,000,000 mark this year. Thanks to the efforts of many, we have a thriving organization that serves Barth families' needs in many different ways. BSF dreams big and is not limited by what seems possible in light of current circumstances.

A by-product of our evolution is the fact that, because of term limits, our current Board of Directors no longer contains any of the original Board members. And while we are still adjusting to this new normal, I believe the organization is as strong as ever. We have assembled a great staff who keeps everything running smoothly without Board micromanagement, and we have terrific Board members who bring incredible intelligence, experience and judgment to our decision-making. Our most recent addition to the Board is Kevin Woodward, a committed Barth Dad and project management professional who has already contributed in many ways to our cause. I look forward to working with Kevin and the rest of the Board, and to more new Board members joining our ranks, to take us to the next level and even greater achievements in the coming years.

One source of reassurance as BSF evolves is that our long-time members remain as committed as ever to our shared mission. Kate McCurdy and Sue Wilkins are the latest and last of the founding Board members to serve out their Board terms, and I would be remiss if I didn't recognize their immeasurable contributions to this organization. BSF has as its core a committed group of Barth Moms, and Kate and Sue are the epitome of what this organization and these Moms are all about. No words can thank these two amazing women enough for what they've done for the organization. Nothing I could write could be as eloquent as the thank-you note their sons Will and John wrote to them on the BSF listserv or the tributes found later in this newsletter. So I will simply add my thanks to Kate and Sue and look forward to their continued and substantial involvement with BSF.

Kate and Sue and the rest of our former and current Board members know one thing — we're all in this together. The oft-repeated saying is that "none of us would have chosen that our son/brother/grandson has Barth syndrome, but if he is going to have a rare disorder, we sure are glad it is this disorder because *this organization* exists to help him." *This organization* is us, a diverse group of people who together can make a much greater difference than any of us can do individually. Each of us, and the organization, will face challenges along the way. We may have to step out of our comfort zones, step up to new challenges, and adjust to new circumstances. But as long as we keep our Barth boys and men in mind, and focus our efforts on their best interests, we will continue to make good progress toward our goals. Thank you all for your support.

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Enthusiasm for Barth Syndrome 2014 Conference

"I feel very fortunate to work with the Barth Syndrome Foundation and the unique and enthusiastic families it serves.

I look forward to future meetings, scientific and clinical collaborations, and meeting new friends." ~ Hilary Vernon, MD, PhD

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Drs. Hilary Vernon and Cliff Takemoto
at the Barth Syndrome Clinic at KKI.²⁰¹²

The 2012 BSF meeting was also a wonderful opportunity to get to know families, clinicians and researchers involved with Barth syndrome. My husband and 6-year-old daughter also thoroughly enjoyed getting to know the community and attending all of the social events. It was very special to have the opportunity for my family to learn about BSF.

My involvement with this special community has not been limited to research. In September of 2012, Dr. Richard Kelley and I co-founded the Barth Syndrome Interdisciplinary Clinic at the Kennedy Krieger Institute. We, along with a team of dedicated clinicians in various specialties, have seen more than 20 families affected with Barth syndrome.

One of our goals is to gather and disseminate clinically useful information in order to better inform the wider medical community about signs, symptoms, and treatment of Barth syndrome. As part of this goal, a Gene Reviews article is in press co-authored by myself, cardiologist Dr. Reid Thompson, and biochemical genetics trainee Dr. Carlos Ferreira. Gene Reviews are often a first go-to for clinicians looking for information about genetic conditions, and we anticipate that the publication of this resource will relieve some of the burden on families in working with clinicians not immediately familiar with Barth syndrome.

I feel very fortunate to work with the Barth Syndrome Foundation and the unique and enthusiastic families it serves. I look forward to future meetings, scientific and clinical collaborations, and meeting new friends.

BSF Seed Funding Leads to NIH Grants

(Cont'd from page 1)

From studies funded in part by the R21 grant, we discovered that cardiolipin-deficient yeast cells exhibited defects in both iron homeostasis and metabolism, as well as defective import of proteins into mitochondria. BSF grants enabled us to explore the role of cardiolipin in these essential functions. We determined that the loss of cardiolipin led to perturbation of iron homeostasis due to defective synthesis of iron-sulfur (Fe-S) clusters. Fe-S clusters are important cofactors in many essential metabolic reactions, including those of the tricarboxylic acid (TCA) cycle, the metabolic pathway that is crucial for basal cellular metabolism. The TCA cycle is essential for cardiac production of ATP, the primary source of energy for contraction and iron homeostasis in the heart. The protein import defect suggested a potential mechanism for perturbation of Fe-S biogenesis. We hypothesized that Fe-S synthesis may be perturbed because proteins required for this function are not properly imported into mitochondria. To carry out studies to test this hypothesis, we were recently awarded an NIH grant (2014-2018) entitled "**The Role of Cardiolipin in the TCA Cycle: Implications for Barth Syndrome.**" There is no question that BSF support has led directly to this NIH grant.

In addition to our research to elucidate the role of cardiolipin in basal cellular metabolism, we wish to determine if the deleterious effects of *tafazzin* deficiency can be reversed by inhibiting the cardiolipin remodeling pathway. Cardiolipin is remodeled in two steps. First, a phospholipase enzyme removes a fatty acid from 'unremodeled' cardiolipin (which has four fatty acids) to form MLCL (a cardiolipin molecule with three fatty acids). Second, *tafazzin* adds a fatty acid to MLCL to form 'remodeled' cardiolipin. The loss of *tafazzin* in Barth syndrome leads to an increase in the MLCL/cardiolipin ratio (which is thought to be a reliable marker for the disorder) as well as to a deficiency in remodeled cardiolipin. Because remodeled cardiolipin has primarily unsaturated fatty acids, the pathology in Barth syndrome has been attributed to the observed deficiency in remodeled (unsaturated) cardiolipin. However, very recently, both Dr. Steven Claypool's laboratory and my laboratory have reported that a deficiency in remodeled cardiolipin is not harmful to yeast cells, and that cells that lack the phospholipase grow normally (Baile et al., 2014; Ye et al., 2014). Intriguingly, deleting the phospholipase rescues growth defects of the *tafazzin* mutant. This suggests that *tafazzin*-deficient yeast cells are defective because the phospholipase depletes cardiolipin and/or increases MLCL levels, not because cells lack remodeled (unsaturated) cardiolipin. Drs. Mindong Ren and Michael Schlame first suggested this possibility in a study published in 2009 showing that inactivating the phospholipase iPLA2-VIA rescues sterility in *tafazzin*-deficient *Drosophila* (Malhotra et al., 2009). We wish to determine if inhibiting

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cardiolipin-specific phospholipases will rescue defects in Barth syndrome cells. The major obstacle to testing this possibility is that human cells have tens of phospholipases, many of which can potentially act on cardiolipin (in contrast to yeast cells which have only one cardiolipin-specific phospholipase). Determining which of these specifically act on cardiolipin is a daunting task that cannot be easily undertaken in mammalian cells. To address this problem, we have developed a yeast bioassay to identify the human cardiolipin phospholipases. The bioassay is based on the hypothesis that expression of a human cardiolipin phospholipase in yeast *tafazzin* mutant cells will be deleterious to these cells, resulting in poor growth of the yeast cells that can be easily observed on Petri dishes. These phospholipases may be potential new therapeutic targets for treatment. The BSF grant that we received this year for **"Identification of Human CL Phospholipases that are Deleterious to *Tafazzin*-Deficient Cells"** will support this project.

I am very grateful for BSF support, which has enabled my research group to continue to develop and test hypotheses relevant to Barth syndrome. It is my fervent hope that our studies will contribute to our understanding of the pathology in Barth syndrome, and to the development of potential treatments for this disorder.

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"I am very grateful for BSF support, which has enabled my research group to continue to develop and test hypotheses relevant to Barth syndrome. It is my fervent hope that our studies will contribute to our understanding of the pathology in Barth syndrome, and to the development of potential treatments for this disorder."

~ Miriam L. Greenberg, PhD



Dr. Miriam Greenberg (center) with her team of researchers at the Detroit Institute of Arts.

7th International Barth Syndrome Scientific, Medical & Family Conference

Make a Difference!

June 23-28, 2014 ~ Clearwater, Florida

A block of rooms has been reserved for our group with a reduced rate of US \$130 per night. The special room rate will be available until May 23, 2014 or until the group block is sold-out, whichever comes first. These rates have also been extended for those arriving early and/or departing later.

1. Booking a hotel reservation is simple. Go to the Hilton's website and click on, "Book a Room" to receive the preferred rate. You may also call 800-753-3954. Please make any arrangements for special needs directly with the hotel at the time of booking. Link to book online: http://www.hilton.com/en/hi/groups/personalized/P/PIECBHF-BARTH-20140619/index.jhtml?WT.mc_id=POG
2. In addition to making your hotel reservation, you will need to register for this Conference with the Barth Syndrome Foundation (BSF). Registration is available on BSF's website. To register with BSF go to <http://barthsyndrome.org/science--medicine/-scientific,-medical--family-conference>.

For assistance or further information, please contact us at bsfinfo@barthsyndrome.org.

Date	Science & Medicine	Family
Monday, June 23		Registration & Welcome Reception
Tuesday, June 24	Patient Discussions (by invitation)	Discussions with BTHS experts and Research Studies (formerly called "Clinics")
Wednesday, June 25	Patient Discussions (by invitation) Physician/Scientist Registration Informal Reception	Discussions with BTHS experts and Research Studies (formerly called "Clinics")
Thursday, June 26	Scientific/Medical Sessions Keynote Speaker Poster Session (Thursday)	Family/BTHS Individual/Sibling Sessions Keynote Speaker Poster Session (Thursday)
Friday, June 27	Scientific/Medical Sessions Varner Award Luncheon Social Event	Family/BTHS Individual/Sibling Sessions Varner Award Luncheon Social Event
Saturday, June 28	SMAB Meeting (by invitation) Closing Ceremony	Breakouts Closing Ceremony

Keynote Speaker for Barth Syndrome 2014 Conference



Dr. Barry Byrne

Barry J. Byrne, MD, PhD — Associate Chair and Professor of Pediatrics and Molecular Genetics and Microbiology, College of Medicine, Department of Pediatrics; Molecular Genetics and Microbiology; Director of the Powell Gene Therapy Center at the University of Florida, Gainesville, FL, USA

The stellar physician-scientist, Barry J. Byrne, MD, PhD will be the keynote speaker at the 7th International Scientific, Medical & Family Conference on Barth syndrome. Renowned as a pediatric cardiologist and for his efforts to find treatments for several rare diseases, Dr. Byrne is a major voice and champion for that underserved community. The Barth Syndrome Foundation (BSF) is fortunate to count on him as a close friend and an important advisor over the years. He served on the Scientific and Medical Advisory Board of BSF, has received a research grant from BSF, has cared for many Barth syndrome patients, and has been a defining presence at the BSF biennial conferences. Dr. Byrne's laboratory is focused on molecular approaches to diagnosis and treatment of heart failure in infants and children which includes Barth syndrome. Dr. Byrne studies glycogen storage diseases (Pompe disease), muscular dystrophies (Duchenne), hemophilias, as well as Barth syndrome, where he uses viral vectors (genetic therapy) in conjunction with stem cells to repair damaged hearts. These

programs are supported by many prestigious organizations like the American Heart Association, Muscular Dystrophy Association, and the National Institutes of Health (NHLBI, NIDDK, and NCCR). He is frequently called upon to advise the NIH. Dr. Byrne was one of the first researchers to publish on his work with the mouse model of Barth syndrome for which he received a BSF Research Grant in 2010. Most importantly, Dr. Byrne is a wonderful physician who gives of himself to the individuals he cares for — in fact, he even climbs mountains for them! We are very excited to hear "Barry" as he tells us about the "shape of things to come" in molecular medicine.

2013 Research Grant Program is the Most Competitive Ever

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

With the completion of the 2013 Barth Syndrome Foundation (BSF) Research Grant Cycle, 12 annual award cycles have committed a total \$3.0 million through 78 research grants to 46 principal investigators. As with all BSF grant cycles, the 2013 cycle grants were awarded the following year, thus being included in 2014 fiscal year expenses. The BSF Board of Directors, with the advice of its International Scientific and Medical Advisory Board (SMAB), and with support from international affiliates, awarded six research projects. Starting in 2013, BSF awarded 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. This upgrade to the BSF Grant Program attracted the largest number of applications which also made the 2013 cycle the most competitive ever.

The BSF Research Grant Program has resulted in many publications which further describe scientific and medical components of this multi-faceted disease and are leading towards new ideas for treatment. A complete list of all grant awardees can be found on BSF's website at www.barthsyndrome.org, and those awarded in the 2013 cycle are featured below.



William T. Pu, MD, Associate Professor, Boston Children's Hospital; Boston, MA

Reactive oxygen species and mitochondrial dynamics in the pathogenesis of Barth syndrome

Award—US \$100,000 over 2-year period

Using iPS cells to understand the pathophysiology of *tafazzin* dysfunction and to screen for potential therapeutic compounds. Dr. Pu has developed induced pluripotent stem cells (iPS cells) from skin cells taken from two Barth syndrome individuals and uses them to analyze the biochemical and metabolic abnormalities at the cellular and subcellular levels. This system uses a state-of-the-art analysis to monitor mitochondrial functions in fine detail and is at the leading edge of research. Building on his earlier work, Dr. Pu will examine how reactive oxygen species (ROS) can lead to mitochondrial defects and damage (this has been implicated by the work of several other researchers) and how the fusion and fission of

mitochondria may be altered in Barth syndrome cells (a new insight). He will use specific chemical antioxidants to reduce ROS and monitor for effect. In the mitochondria fusion-fission studies, he will use live cell imaging with fluorescent compounds to visualize this process as well as monitoring OPA1 expression which is a protein directly involved with mitochondrial fusion. In addition, Dr. Pu will continue to screen potential therapeutic compounds using his iPS cell system and characterize their value for treatment. These mechanistic studies are being performed in an elegant cell culture model of Barth syndrome using a multiplicity of sophisticated assays to assess interventions that may be applicable to Barth syndrome individuals.



Miriam Greenberg, PhD, Professor and Associate Dean, Wayne State University; Detroit, MI

Identification of human cardiolipin phospholipases that are deleterious to *tafazzin*-deficient cells

Award—US \$50,000 over 1-year period

Using yeast as a model to identify which human phospholipase(s) can alter *tafazzin* dysfunction. Dr. Mindong Ren and colleagues showed that some of the traits of the fruit fly model of Barth syndrome could be reversed by inhibiting a phospholipase that is involved with cardiolipin processing. Cardiolipin is a fat-like substance that is altered in Barth syndrome. Phospholipases are enzymatic proteins that alter certain types of fat-like substances in the cell. Dr. Greenberg discovered that deleting the only phospholipase gene found in yeast (*cld1*) improves the growth of the *tafazzin*-deleted yeast cell line. Building on the idea that phospholipase inhibition may reverse *tafazzin* deficiency, Dr. Greenberg is setting

up a screen in yeast cells to test human phospholipases for their effects on the *tafazzin* deletion yeast mutant with the expectation that several of these phospholipases may work just like the one identified earlier by Dr. Ren and colleagues. If positive, inhibition of human phospholipase activity may be therapeutic and may lead to a new drug-discovery project. In aim 1, Dr. Greenberg will develop the yeast bioassay using the human phospholipase gene that Dr. Ren had identified and the *tafazzin* plus *cld1* double deletion yeast mutant grown under conditions that require mitochondrial function. In aim 2, she will clone the dozens of human phospholipases that are known from the human genome project and determine which ones also impair growth of the yeast model system. Dr. Greenberg will confirm any positive results from aims 1 and 2 using Barth syndrome lymphoblasts treated with specific inhibitors of the lipase (RNAi) under study. The next step would be to find pharmaceutical phospholipase inhibitors that can work in the same manner as a potential therapy for Barth syndrome.

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2013 Research Grant Program is the Most Competitive Ever

(Cont'd from page 7)



Douglas Strathdee, PhD, Head of Transgenic Technology, Beatson Institute for Cancer Research; Glasgow, Scotland
Characterization of a conditional knockout of *tafazzin* in the mouse

Award—US \$49,837 over 2-year period (Funds provided by Barth Syndrome Trust, UK)

Making a conditional knockout mouse model of Barth syndrome that can delete *tafazzin* in certain organs. Many previous attempts to make a knockout mouse model of Barth syndrome have been frustrated for reasons that are unclear. A knockout mouse model is one where the entire gene is deleted from the mouse genome. Dr. Strathdee has succeeded in making a mouse that can genetically transmit (germline transmission) a modified *tafazzin* gene (floxed construct) which has never been reported before. This modification is able to cause the deletion of the *tafazzin* gene when the mouse is mated with special Cre-recombinase containing mice that are available from other researchers. This system will allow mice to be

totally deficient in the *tafazzin* gene which is different from the knockdown mouse model currently in use. Dr. Strathdee will determine if his knockout mouse model has characteristics that resemble the symptoms suffered by Barth syndrome individuals. In addition, this mouse model will allow the *tafazzin* gene to be deleted at certain times in the development of the mouse or deleted within certain organs of the mouse. This “conditional” knockout property of his mouse model will allow researchers to see how *tafazzin* deficiency impacts individual organs or organ systems, and most importantly how it impacts the heart. Dr. Strathdee will also breed his knockout mouse line with different genetic strains of mice to determine if the symptoms can be altered by genetic “modifiers” present in those other strains of mice. Dr. Strathdee has also uncovered evidence that the difficulties that many laboratories previously encountered in trying to make a *tafazzin* knockout mouse model probably were the result of the placenta being negatively affected by the loss/modification of the *tafazzin* gene.



Stacey Reynolds, PhD, OTR/L, Associate Professor, Virginia Commonwealth University; Richmond, VA

A systematic investigation into sensory and motor based feeding issues in boys with Barth syndrome

Award—US \$18,732 over 1-year period (Funds provided by Association Barth France)

Investigating taste and sensory issues with Barth syndrome individuals. Dr. Reynolds will expand upon her taste-testing data, originally obtained from Barth syndrome individuals at the 2012 Conference, by including for the 2014 Conference: propylthiouracil (PROP) tasting (a chemical known to vary in its ability to be tasted), determining the density of tongue tip papilla analysis by photography, salt preferences, food inventory, and a parental questionnaire about their son’s behavior. This work builds on her 2012 publication and on the data collected at the 2012 Conference. Dr. Reynolds also plans to collect home videos to evaluate early sensory and motor features of Barth syndrome which may allow her to include young children who otherwise could not be part of the study. Dr. Reynolds also plans to engage volunteer members of the BSF community to translate this knowledge back to the community and encourage the

clinical research process in general. This is consistent with Dr. Reynolds’ long-term goal to develop therapeutic approaches to evaluate and treat sensory and motor-based feeding issues in boys/young men with Barth syndrome.



Grant Hatch, PhD, Professor, University of Manitoba; Winnipeg, Manitoba, Canada

Tafazzin knockdown alters hepatic lipid metabolism

Award—US \$49,995 over 1-year period (Partial funding provided by BSF of Canada)

Investigating liver metabolism (fat) in knockdown mice as part of the Barth syndrome pathophysiology. Dr. Hatch has shown that the knockdown mouse model of Barth syndrome parallels the human condition (“skinny” phenotype) by demonstrating that the mice weigh less than their wild type counterparts particularly in the fat pad areas or fat depots and in the amount of fat in the liver. Dr. Hatch will study the physiology of the knockdown mouse model from the perspective of lipid metabolism using respiratory exchange ratio analysis, heat production, plasma lipid analysis, and activity levels. He has focused on understanding how the fat depots in this mouse model are different, and how this produces the “skinny” phenotype which distinguishes them from the wild type mice. Lipid or fat metabolism in any animal is

greatly influenced by the liver, however the liver has not figured prominently in our understanding of Barth syndrome. For example, plasma lipid (fat) levels in Barth syndrome individuals are only slightly altered. By studying the knockdown mouse model of Barth syndrome, the contribution of liver metabolism to the pathophysiology of Barth syndrome will be systematically studied with the goal of improving treatment.

(Cont'd on page 9)



Nathan N. Alder, PhD, Assistant Professor, University of Connecticut; Storrs, CT

Investigation of cardiolipin-dependent respiratory complex activity and development of small molecule lipid analogs

Award—US \$50,000 over 1-year period

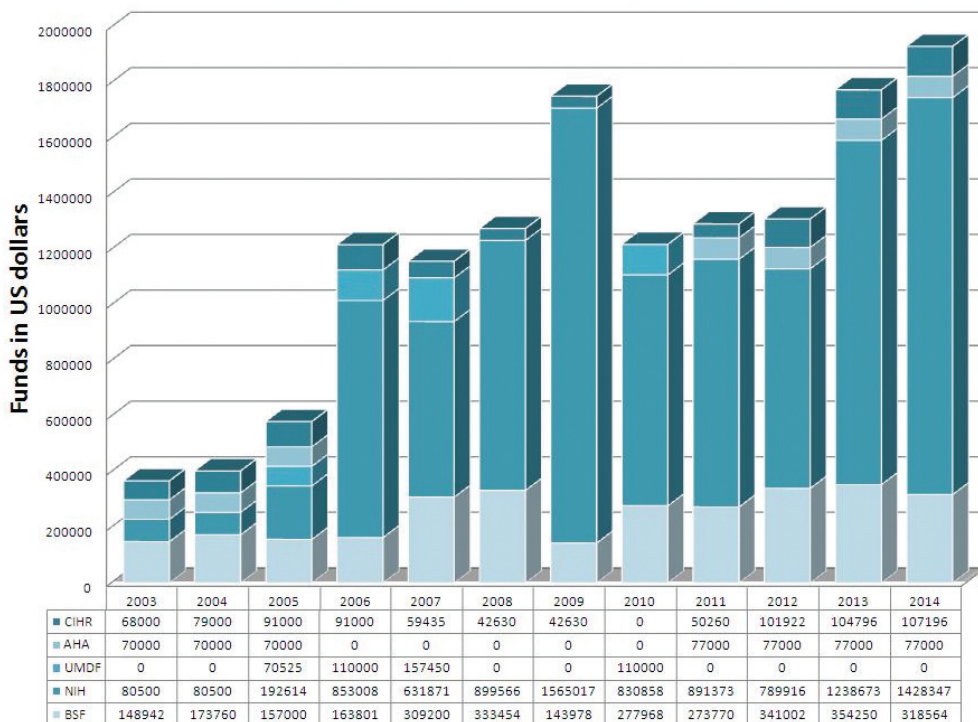
Using nanodisks and nanoparticles made with different cardiolipin molecules to examine mitochondrial energy structures (complex IV). Dr. Alder has developed a methodology to study the biophysical assembly of protein complexes (electron transport chain or ETC) that make up part of the machinery which mitochondria use to produce energy in the cell (OXPHOS). His system, which involves making synthetic nanodisks and nanoparticles, is novel and could be superior to methods now used to prepare synthetic membranes (detergent solubilization of organelles). His technology will allow the direct testing of how

different lipids (such as the Barth syndrome-specific lipid MLCL) affect the ETC (specifically, complex IV). This unique system will allow experiments to be carried out that are directly testable and have more relevance to the issue of how cardiolipin abnormalities affect the function of mitochondria. Cardiolipin is the fat-like substance that is altered in Barth syndrome. Dr. Alder will use potentiometric and spectroscopic techniques to determine the effects of modified cardiolipin molecules on complex IV activity and properties.

Summary

Our research grants have evolved over the years from simply looking at basic scientific questions to now evaluating potential therapies for Barth syndrome. The graph below illustrates the momentum and progress we've made, both in terms of the amount of funding that BSF has been able to provide to research as well as the amount of additional research that is now being done without direct funding from BSF. The increased awareness and interest in Barth syndrome research led to a record number of grant applications received by the organization this past year. While much work remains to be done, we believe our strategy of "seeding" research interest in areas implicated by Barth syndrome is paying significant dividends.

Funding Sources For Barth Syndrome Research



Note:

- The Y axis is in US dollar equivalents
- CIHR = Canadian Institutes of Health Research
- AHA = American Heart Association
- UMDF = United Mitochondrial Disease Foundation
- NIH = National Institutes of Health
- BSF = Barth Syndrome Foundation & Affiliates

BSF SMAB Welcomes Dr. Marc Tarnopolsky

By Kate McCurdy, Former Board Member; Former Scientific & Medical Advisory Board, ex-officio; Barth Syndrome Foundation

It is with great pleasure that the Barth Syndrome Foundation (BSF) Board of Directors welcomes a new member of our International Scientific and Medical Advisory Board (SMAB).



Mark Tarnopolsky, MD, PhD is an expert in mitochondrial and muscle disorders and is very engaged in both clinical practice and research. He is quite familiar with BSF as he delivered an excellent presentation at the Scientific and Medical sessions of our 2010 International Barth Syndrome Conference and also has served several times as an outside reviewer for various grant applications submitted to BSF.

Dr. Tarnopolsky holds an MD with a concentration in Neurology and Physical Medicine & Rehabilitation and also has a PhD in Cell Biology and Metabolism. He is a Professor at McMaster University in Ontario, Canada and heads the Neuromuscular and Neurometabolic Disorders Division of Pediatrics there. We are sure he will have much to contribute to our group, especially as we continue to focus on advancing treatments for Barth syndrome. We are delighted that he now has joined us as an official advisor.

In addition, we are very pleased to report that each of the five members of our SMAB whose terms were up at the end of 2013 have agreed to serve for another four-year term. We are very fortunate to have Miriam Greenberg PhD, Grant Hatch PhD, Colin Steward PhD FRCP FRCPCH, Jeffrey Towbin MD, and Ronald Wanders PhD continue as SMAB members.



Dr. Miriam Greenberg



Dr. Grant Hatch



Dr. Colin Steward



Dr. Jeffrey Towbin



Dr. Ronald Wanders

Kate McCurdy Changes Roles

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



Kate and Matt Manning BSF's booth 2006

Kate McCurdy is responsible for establishing the Science and Medicine Program of the Barth Syndrome Foundation (BSF); an outstanding program which I inherited. Kate recognized early on the need for a group of scientific and clinical experts to help BSF accomplish its goals, namely, to foster a better understanding of Barth syndrome and to find a treatment. Starting from a blank sheet of paper, she assembled the Scientific and Medical Advisory Board of the BSF (SMAB) to guide BSF in encouraging Barth syndrome research and researchers. Kate also established the competitive BSF Research Grant Program which has been the major influence to research and researchers over the years. One of the top priorities of the SMAB is to evaluate the Research Grant Program applications each year. Many of the advancements in understanding Barth syndrome have come as a result of the Research Grant Program that Kate initiated and sustained, and which has just completed its 12th cycle. She has been a tireless advocate for Barth syndrome research and researchers and significantly increased BSF's profile in the scientific and medical community.



Cathy Ritter and Chris Hope 2013

Now, due to the institution of term limits, Kate is stepping down from the BSF Board of Directors and therefore will also rotate off the SMAB. She will turn over her duties as an *ex-officio* SMAB member to the capable stewardship of Cathy Ritter. As a BSF Board member, Cathy will serve on the SMAB and assume the responsibilities of liaison between the BSF Board and the SMAB. As a Registered Nurse now working as a transplant coordinator, a Barth mother, a member of the BSF Canada Board and a member of the BSF Board, Cathy brings a wealth of experience and expertise to this role. We are very excited to add her to this already strong team.

Fortunately, we can still count on Kate to be available to provide her good counsel and guidance as the BSF Science and Medicine Program reaches towards its goals. Kate, thank you for all your wonderful work over the years!

Sue Wilkins Steps Off Board

By Shelley Bowen, Director, Family Services & Awareness, Barth Syndrome Foundation



(L-R) Shelley and Sue ²⁰¹²

I met Sue Wilkins in 1998. From the moment I first spoke to her, I knew I had met a kindred spirit in more ways than one. When Sue, Anna and I first met, we knew talking about Barth syndrome wasn't enough: we had to do something about it.

Our first task was to bring everyone who knew anything about Barth syndrome together. Sue recruited Dr. Kelley to organize the first scientific and medical gathering of experts on Barth syndrome. She secured the Second Presbyterian Church as the venue for our family meetings.

Then, Sue thought it would be nice to do a little something extra while we were all together, so she called upon a friend who donated baseball tickets for the families, which happened to be located in the President's Suite at Camden Yards. Sue secured our first donation to help fund the meeting. The remaining funds were used as the seed money to establish the Barth Syndrome Foundation (BSF).

Thank You, Steve Groft!

By Kate McCurdy, Former Board Member; Former Scientific & Medical Advisory Board, ex-officio; Barth Syndrome Foundation



Stephen C. Groft, Pharm. D. retired as Director of the Office of Rare Diseases Research at the US National Institutes of Health (NIH) earlier this year, and we at Barth Syndrome Foundation (BSF) want to add our thanks and congratulations to this wonderful man who has done so much personally for us and for all those with rare diseases.

Since BSF began, Dr. Groft has been a sage advisor and supporter of our foundation. His career is impressive and one which shows why he is considered the father of rare disease policy in the US. During the last 30+ years, Dr. Groft has worked at the US Food and Drug Administration (FDA) in the Office of Orphan Products Development, at the US Department of Health and Human Services (HHS) as Executive Director of the National Commission on Orphan Diseases, and at the NIH since 1989 when it established its first Office of Rare Diseases Research. He always has fought for the cause of rare diseases, including early work leading up to the passage of the Orphan Drug Act of 1983, which made it more attractive for pharmaceutical companies to invest in the creation of drugs that benefit those with rare (or "orphan") diseases.

In the Fall of 2000, just after BSF had been incorporated, the five original BSF Board members first gathered to discuss what BSF's mission and specific goals should be. We knew we had a daunting task ahead when we decided that we wanted to facilitate and accelerate Barth syndrome research. Dr. Groft was one of the primary people who helped us as lay advocates, who were ourselves neither scientists nor doctors, figure out how we could have a positive impact in this arena. He has been our champion from the beginning, giving us advice,

Sue was a founding board member of BSF and served as our very first treasurer. Sue continues to raise funds for BSF. She has been a steady influence on all of us over the years; she is well known to our donors for her truly exemplary, personal and heartfelt thank you letters that she has sent over the years. In fact, one donor once told us, "Sue's thank you note made me break out my checkbook and donate again."

Sue's encouraging spirit and profound kindness has endeared her to all of our families. Her arms have given many embraces, her hands have gently enveloped the hands of many concerned mothers, her experience has given much comfort and her smile has reminded us things were going to be alright. And most importantly, her dedication to walk the walk and not talk the talk has inspired our confidence in her.

Due to term limits, Sue's service as a board member ended this month. However, her devotion to our families and dedication to the mission of this organization has not. To some, Sue's departure from the board may be considered the end of a chapter of BSF history. However, Sue would be the first to remind all of us that this is the beginning of a new era.

letting us know about resources that were available, including us in various NIH activities and meetings, offering us contacts and helping us win financial support for our conferences. And he has known about a number of our Barth guys by name and often would ask how they were doing. We were particularly delighted when he helped kick off our very first BSF conference in 2002 and again when he agreed to come back a decade later as our keynote speaker in June 2012.

There are over 6,500 rare diseases that affect as many as 25 million people in the US alone. By both advancing rare disease research and by giving patients, their families and other advocates a voice, Dr. Groft has made a huge difference in the lives of these people and many more around the world. His quiet but determined manner, his deep and sincere compassion, his integrity and his creativity have helped him lay an amazing foundation for even greater things ahead in rare diseases, and we will continue for a long time to think of him when good things happen. The rare disease community, including those of us at BSF, has benefitted greatly from Dr. Groft's unwavering devotion to our shared cause.

As BSF Science Director Dr. Matt Toth stated, "He and his wonderful staff have always been a resource and a friendly place where BSF and similar patient interest organizations can turn. The Global Rare Diseases Patient Registry and Data Repository (GRDR) is just the latest advancement, and I truly believe it will be a vital part of what we all want to see accomplished – a world where there are treatments or hope for treatments for rare diseases."

Dr. Groft's fingerprints are all over what we have accomplished as a patient organization; he helped us chart our pathway forward and gave us hope. We are profoundly grateful.

Donors Are Vital Members of Our Family

By Stephen B. McCurdy, Chairman Emeritus, Barth Syndrome Foundation



Researchers collaborate at BSF's 2012 Conference.
(Photo courtesy of BSF 2012)

I wish our donors could come to our BSF Conference!

Our scientists and researchers come and every time they meet, the intellectual excitement is palpable! Our families come — more and more each conference — and every meeting strengthens their friendships, their ability to care for their kids and their mutual devotion and optimism. Our Board, staff and volunteers come and see the amazing results of all their hard work, organization and love. The Barth Syndrome Foundation is an astonishing thing. We have no headquarters — everyone works from their home. The entire thing functions at a distance on the internet except when a few folks gather at board meetings or clinics or regional outreach meetings. But once every two years, everybody gathers at the Conference and renews their personal inventories of knowledge, plans, optimism, ideas, excitement, love and energy.

It's what happens when people become devoted to a single cause...a simple cause...saving children they know personally.

That is the amazing thing that our donors know without ever coming to our Conference, without attending a single scientific session or holding the hand of a boy having blood drawn at a clinic. They get it because they have come to know our boys. Our earliest and most devoted donors knew and loved our families and their boys personally long before they knew about Barth syndrome...no internet required! But over the years, as a few families were willing to have their stories told more publicly in places like: Reader's Digest, The Discovery Health Channel, The Today Show, the Associated Press and many local newspapers and TV broadcasts, and yes, over the internet, more and more donors have been attracted to our progress and our cause. They know our story even when they don't know the boys themselves.

Today, many of you reading this will, indeed, be devoted, repeat donors. You provide the fuel that enables our growth and our continued success. As you can read elsewhere in this newsletter, with progress comes the need for more financial resources. When we started this foundation, we didn't have enough money for chairs or tables in the hotel meeting room where we met. Since then, your generosity has enabled scientists to create yeast, zebrafish, fruit fly and mouse models of Barth syndrome, that accelerate research. And, recently, scientists created human stem cells that carry the Barth gene, another giant leap ahead in our search for a deeper understanding of what causes Barth syndrome and ultimately, the quest to find effective treatments and a cure for this too-often deadly and disabling disorder.

The upcoming Conference in June will be especially exciting. The scientists have a lot of new progress to share and discuss and more families than ever will be in attendance. We all know that none of us would be there without your faithful support and devotion to our cause. We are enormously grateful. We wish you could be there with us. We will let you know what happens, and we promise to take pictures.



(L-R) Megan, Grady, Henry (age 2) and John.
(Photo courtesy of Henry's Family 2014)

Fundraising and Awareness Events

Henry Heart Walk

Henry was a San Francisco Bay area toddler when he was diagnosed with Barth syndrome. To increase awareness and raise money for BSF, his parents invited friends and family to join Henry for a walk around the local reservoir in February. Nearly 150 participants donned matching red T-shirts to hike the reservoir trail, before enjoying a sausage, hot dog, fruit, and cupcake lunch — some of Henry's favorite food. The walk itself was free. "We pre-sold t-shirts," Megan explained, and the family encouraged donations to the Barth Syndrome Foundation.

"We had great support from local businesses (Peet's Coffee, Susie Cakes, Lafayette Carwash, Boswell's Party Supplies, and Whole Foods, all in Lafayette, plus East Bay Screenprint, Concord, and Gianna's Baking Company, Castroville)," she said. And the local crowd was joined by a group of eight families of friends from Ft. Worth, Texas, wearing the same t-shirts and walking in solidarity with

(Cont'd on page 13)

(Cont'd from page 12)

Henry and a new friend from a nearby community – the only other Bay Area child known to have Barth syndrome. "The past year and a half has been quite a journey, definitely the most trying time in our lives, but also a time of feeling very loved and supported by our family, friends, and community," Megan said. "We're very thankful."

Conference Support

Many of our families are searching for a few good sponsors. The BSF Conference is coming up in June (23-28) and will be held in Clearwater, Florida at the Hilton Clearwater Beach Hotel. There are opportunities to sponsor meals, events, presentations – over 15 sponsorship opportunities ranging from US \$50 to US \$10,000. The Wilkins, the Woodwards and the Marra-McCormacks are all approaching their local businesses and employers in Nebraska, Maryland and Massachusetts to ask for their support of this important and exciting event. We know we haven't covered all the states and communities where our families live and we are looking for more help in gaining support for the Conference. If you would like to help, please contact Sandra Stevens, Fundraising Project Manager at sandra.stevens@barthsyndrome.org.

Honoraria and Memorials

Every year, we receive donations in honor of heroes — past and present. Most of these gifts are in honor of our own Barth boys and young men, but many are in memory of loved ones recently deceased. Newly married couples have asked that donations be made to BSF. Those having a Bar or Bat Mitzva also ask guests to remember BSF. It's clear that the Barth community is growing in leaps and bounds and has earned a place of respect and appreciation to be included in so many personal, important family celebrations and gatherings. We are honored to be remembered and grateful to be included.



Ghent Lummis, member of Team Will.
(Photo courtesy of Ghent Lummis 2013)

Ironman

Ghent Lummis, one of our veteran Team Will Ironmen from Houston, Texas, is taking on the Lanzarote Canary Island Ironman race on May 17th. A full Ironman race, Lanzarote is reputed to be the toughest Ironman in the world with high winds and over 2,551 meters of climbing in the bike leg. Ghent is dedicating his race to BSF and raising money on the internet. He will also attend the BSF Conference in June to cheer on our Barth guys as they complete their exercises as a part of the clinic.

Coach Gary Rodbell will lead Team Will to compete again in the Jarden Westchester Triathlon on September 21st in Rye, NY. This event, a "sprint triathlon" ("only" a .9 mile swim, 26 mile bike ride and 6.2 mile run!) attracts individuals and relay teams who all work equally hard raising money for BSF and training to achieve a personal best in the race. It's a fun event followed by an afternoon cookout at the McCurdy's home to rest and celebrate.

Stefan Tunguz, another Team Will Ironman from Larchmont, NY, is entered in the Ironman World Championship in Kona, Hawaii on October 11th. Stefan will

be wearing the Barth colors, and he and his wife, Julie, have graciously agreed to work with BSF staff to use this event to raise money and awareness for BSF. The race will certainly be broadcast on TV, so look for Stefan!

Year-End Giving

Most charities raise more than half of their annual budgets in the last two months of the year, and BSF is no different. It is always a worrisome time of year for Lindsay and the Board, as they watch to see if enough donations come in before New Year's Eve to cover the expenses that have been incurred over the previous 12 months. Most of the time, we make it. However, we have come up short a couple of years. This is a particularly challenging year for two reasons. First, it is a Conference year which increases expenses. Second, for the first time ever, we are actively pursuing a drug trial with the FDA — a very expensive process but one which we are very eager to support! Barth families, we will need as many of you as possible to help with year-end fundraising — writing letters, encouraging family and friends to remember to give to BSF. Don't forget to start pulling your holiday card list together and confirming addresses. Let's all send "Henry" Cards to all of our friends, colleagues and family this year. And donors, please be as generous as you possibly can. We promise to use your donations in ways that will efficiently and effectively push us forward toward our shared goals.



Don't forget to place your order for your Henry cards!

Meet Sandra ~ Working with us toward a Cure

By Sandra Stevens, Fundraising Project Manager, Barth Syndrome Foundation



(L-R) Lindsay Groff and Sandra Stevens
at Indiana School of Philanthropy.
(Photo courtesy of BSF 2014)

I can't tell you how excited I am to be able to serve you. As Lindsay said in her article, the Barth Syndrome Foundation (BSF) is growing, and I'm a direct by-product of that growth! I'm here to help the BSF team increase fundraising to provide increased funds; to expand our research and the standard of care and to facilitate our quest for a cure for this dreadful disorder.

When I first learned about this new position at BSF, I went to the website to discover more. There, I found and watched two videos where parents and affected boys described their journeys, how BSF has helped them, and the importance of hope.

I was instantly hooked on the mission, even before I knew I had the job! That was a powerful introduction to my relationship with BSF...one that I will never forget and one that drives me every day to do my best job possible and fight for these boys and young men.

I'm from Britain, and my background includes time at the British Broadcasting Corporation (BBC) in London, where I was responsible for helping people renew their television license. I believed in the high standard of programming, and it

was a joy for me to communicate the message that if you want quality, you have to invest in it. If you want the best possible solution, you can't shy away from the fact that it costs money. I felt this was a precious piece of learning to bring to my new role at BSF.

You see, if we want to fund groundbreaking research and get valuable information into peoples' hands, it's going to cost money... money that can only come through generous donations from our faithful partners like many of you.

However, this is what I hope you will never forget. You are stakeholders of the Barth Syndrome Foundation. I do not take that lightly. It will be my job to tell you our needs, ask you to support those needs, report back to you on how your gifts are making an impact, and provide you with the best service I can.

We will not let you down.

We've recently invested in some really important tools that will allow us to serve you better. This is all part of that progress we've been talking about.

Now it's time to put those tools to use. Our plan this year is to communicate more frequently with our donors, not only with appeals, but to keep you up-to-date with the difference your support is making. We plan to introduce a monthly giving program to give you a simple way to give regularly. We'll also be reminding you of how Planned Giving can help secure the future of BSF.

And, we'll be harnessing our most valuable asset of all, YOU, our dedicated families, volunteers and donors. So many of you have already raised huge amounts of much needed money with walk-a-thons, raffles, golf tournaments, baseball parties, Ironman races, dance parties, bake sales, bowling parties, fencing tournaments, and more. You are incredible.

At the upcoming conference this year, I hope many of you will be able to attend a special session on Wednesday afternoon dedicated to fundraising. Not only will we give you some great tools, but we'll have fun brainstorming new ideas to raise more money for BSF.

A lot of good work has already been done, and it's my intention to take that good work and continue expanding our efforts...efforts that will have an amazing impact on the boys and young men we all serve and love.

I look forward to talking with and meeting you in the near future. Thank you for welcoming me into the BSF family.

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 **77** 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 Δ). 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
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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
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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
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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
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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.

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
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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.

National Institutes of Health (NIH)

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.

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)



Barth Registry 2.0 Expected to Roll Out in May!

The Barth Registry 2.0 is expected to roll out in May. Please bookmark www.barthsyndrome.org for more information as it becomes available. For those of you who are attending BSF's 2014 Conference there will be an overview provided.

Barth Syndrome News From UK and Europe

Featuring the work of the NHS Barth Syndrome Service and the Barth Syndrome Trust.

News from the Trust

By Michaela Damin, Chair, Barth Syndrome Trust

Barth Syndrome Research in Scotland

We are excited to be funding our first “BSF approved” Barth syndrome research project in the UK. The project “**Characterization of a conditional knockout of tafazzin in the mouse**” is being conducted by Professor Strathdee PhD, Head of Transgenic Technology, Beatson Institute for Cancer Research, Glasgow (see page 8 for more details). We wish Professor Strathdee every success and look forward to being able to give you an update soon.

We are grateful for the generous support of our donors and fundraisers who have provided the \$49,837 (£29,800) for this important project which we believe will have global impact.

Bristol Service and Clinic Update

The annual clinic was held on 8-9th May 2014. Each year, the dedicated Bristol team strive to improve the service provided to families. This year, a special Memory Day meeting on the Friday enabled families to celebrate and remember the lives of the boys who are no longer with us but who remain in our hearts and minds. The Barth Syndrome Trust hosted a fun Family Day at the Bristol Zoo on the Saturday after the clinic and everyone enjoyed the chance to meet outside the hospital environment.

Other news from our Bristol Service team...



Farewell and Thank you, Dr. Tsai-Goodman

By Debbie Riddiford, Barth Syndrome Clinical Nurse Specialist, NHS National Barth Syndrome Service

Dr. Bev Tsai-Goodman has decided to stand down from her post as Cardiologist for the Barth Syndrome Service in Bristol due to her many commitments. We would like to take this opportunity to thank Bev for all her hard work in setting up the nationally commissioned Barth Service and for her ongoing support and dedication. We will miss Bev as a colleague and her daily enthusiasm. We are sure the boys and their families will also miss her being part of their visit to the Bristol clinic.

Welcome Dr. Jonathan Forsey

On the plus side, we would now like to introduce Dr. Jon Forsey who will be continuing Bev’s work. Jon’s major interest is in cardiomyopathy, and he has been busy looking through many of the echocardiograms that Bev and her colleagues have performed in the past.

Development of New Diagnostic Genetic Test

By Dr. Colin Steward, PhD, FRCP, FRCPC, Clinical Lead Consultant, NHS National Barth Syndrome Service

This is not the only change in the NHS service. In the next newsletter, we hope to be able to tell you that we have gone live with a new diagnostic genetic test that Maggie Williams and her colleagues have been developing at Bristol Genetics Laboratory (BGL).

In the past, we have always relied on single gene sequencing of the *TAZ* gene to confirm a diagnosis of Barth syndrome. We would still do this for anyone who has an abnormal blood cardiolipin test, because the cardiolipin test is so rapid, reliable, cheap and easy to perform. However, we quite often receive specimens from boys who have lost their fight with cardiomyopathy, but where no sample is available for cardiolipin testing and their diagnosis is unknown. We have always felt powerless to help families like this if testing turned out to be negative for Barth syndrome mutations; many of you will be aware of just how difficult this is for the families concerned.



(Cont’d on page 19)

This will start to change with the new test which relies on a modern genetic technique called Next Generation Sequencing (NGS). This will allow us to simultaneously sequence 73 different genes which can result in dilated cardiomyopathy in infancy. Many doctors are still unaware of cardiopilin testing, and we also hope that the availability of this test will allow us to identify males with Barth syndrome who have missed being identified so far.

Proposed Drug Trial

By Dr. Colin Steward, PhD, FRCP, FRCPC, Clinical Lead Consultant, NHS National Barth Syndrome Service

"...This just shows what the Barth syndrome community is capable of and provides an inspiration to us all."

Colin Steward, PhD, FRCP, FRCPC

In spring 2013, the NHS Barth Syndrome Service applied to National Institute for Health Research (NIHR) for funding to perform a randomised trial of two drugs, bezafibrate and resveratrol, which had been shown to improve the cardiopilin changes that we see in Barth syndrome cells grown in the laboratory. Discussions between NIHR and the NHS team about funding are still ongoing, and we hope to be able to give you positive news in the next newsletter. This is, however, just one of many steps in the very complicated process involved in setting up a modern drug trial, and we would stress that national regulatory approval (from the Medicines and Healthcare Products Regulatory Agency, MHRA – the UK equivalent of the FDA) and ethical trial approval would still be required. This process could easily take six months from getting news of funding.

The NHS team wish to stress how grateful we are to Dr. Mindong Ren and his scientific colleagues at New York University for performing the laboratory experiments which underpin this application and for being so generous in allowing us to show their data to the NIHR. Also to both Michaela and her colleagues in BST for helping us to develop the plan that we have proposed for this trial and to Dr. Matt Toth for helping us to write the application and helping all the scientists and doctors to work together. This just shows what the Barth syndrome community is capable of and provides an inspiration to us all.

Education

In the last edition of this newsletter, parents wrote about their sons starting secondary school. Here, Gill tells how she addressed one of her main worries when her son, Jack, started primary school.

Germ Warfare in Primary School

By Gill, Mother of Jack, age 7



Jack, age 7, confident at school.
(Photo courtesy of Jack's parents 2014)

One of my main concerns when Jack started school was how I could protect him from all the other germ children! The school staff were very reassuring, but as we all know, classrooms can be rife with bugs and viruses and the pressures on working parents mean sometimes children are sent to school when they would be better kept at home! On top of this, I doubted that other parents would take me seriously. Jack presents as very well to the untrained eye.

Back then, I was still bothered that people may have imagined me to have some weird attention seeking disorder if I made a fuss! Nevertheless, I wrote a positive open letter to the whole school, added Jack's photo, and explained how excited we were to be joining the school. I included the main Barth syndrome symptoms, BST website details and my telephone number with a request that parents let me know if their children had a temperature, sickness or maybe brewing something infectious. The school kindly sent this out before we started. I followed this up with wallet-sized business cards with contact numbers which I gave out to parents of children in Jack's class. It worked really well. Parents would often call me before school so I could decide what to do.

The school was very supportive, and over time my confidence grew and now that Jack is on G-CSF, this has helped a lot.

If I have any advice for parents with boys about to start school it would be don't worry about other people, get the school on your side, be brave and stay in control of what's happening as much as possible.

(Cont'd on page 20)

Barth Syndrome News From UK and Europe

(Cont'd from page 19)

Meeting the Specific Needs of Each Child

By Michaela Damin, Chair, Barth Syndrome Trust

Even two brothers with Barth syndrome can be affected very differently so when it comes to starting pre-school or primary school, work out what specific needs your child has and how best to meet them.

Boys with Barth syndrome can sometimes struggle with gross and/or fine motor skills, fatigue and frequent absence due to illness, tiredness or medical appointments. Most parents strive to make daily life as normal as possible which includes a certain amount of risk taking — we cannot shield our children from every minor illness or accident that is a part of normal social interaction, but we can and should think about ways to ensure that school is a safe environment in which to learn and to have fun with peers.

Please take a look at our Education Guide on the Barth Syndrome Trust website which provides information about various safety issues and adaptations that a young child with Barth syndrome might need when starting primary school. You may also want to use the Care Plan for School template.

And finally... changes to legislation in UK which may well affect you:

In the future, your local council will have to draw up an Education, Health and Care (EHC) plan instead of a statement of Special Educational Needs (SEN), publish a 'local offer' of services and offer a personal budget. Find out more about changes on the Contact a Family website at www.cafamily.org.uk/influencing.

New Disability Benefit in UK

Personal Independence Payment (PIP) is a new disability benefit replacing Disability Living Allowance (DLA) for people aged 16 to 64. DLA continues for children aged under 16. Like DLA, PIP has two parts — the mobility component and the daily living component. For further information about assessment, entitlement to PIP, rates and criteria, please call Contact a Family on their freephone helpline on 0808 808 3555, or email helpline@cafamily.org.uk.

Making the Research Possible

It is wonderful to be able to report significant milestones such as the funding of a research project in the UK. This wouldn't be possible without everyone's fundraising efforts and donations. The first step in the long journey to a cure is fundraising. For news of fundraising events and donations from UK and Europe, please see our fundraising insert.

Memorial Donations

Donations were received in memory of the following boys and other family members:

Terry Farrow

John Skerratt

Jack Reddin (22 December 2009 - 29 December 2009)

Sebastian Vavasour (24 June 2011 - 11 December 2013)



In Loving Memory of Sebastian

By Laura and Joe, Parents of Sebastian



Sebastian
24 June 2011 — 11 December 2013

Born in June 2011, Sebastian arrived with characteristic drama. After the dust settled, we walked through the door of our little house, a family for the first time. All parents know that the story doesn't really end there, but it's true to say that for the first 17 days we had a blissful, sleep-deprived time getting to know each other. Then on 11 July, Sebastian became critically ill.

Sebastian defied the odds, and just a month later we were back at home. Only now we were told that our little boy was in severe heart failure. Alongside feeding and changing Sebastian, we were having to give him drugs around the clock.

Always a little charmer, Sebastian spent the next six months winning the hearts of his nurses, doctors, in fact anybody who he had the chance to meet. His heart became steadily stronger, and by Christmas we were thrilled to be told that he could be expected to make a full recovery.

His medical team continued to reassure us, but we were worried that there was more to Sebastian's story. After a fateful day searching the internet, Laura came upon a rare disease called 'Barth syndrome'. We made the call, and by the following week the diagnosis was made. Sebastian had become a member of an extremely exclusive club!

In September 2012, Sebastian attended his first Barth Syndrome Clinic. Although he really was the baby of the group, the way in which he took to the other boys, and they to him was incredible. We couldn't believe how tender the bond between them all was.

Sebastian's second Christmas was a joyous time, and in February he became a big brother! Greeting Gabriel with 'What's that?' Watching the two of them together was amazing.

Again instinct told us that there was something going wrong, and by May, Sebastian was in intensive care. By June, he had been transferred to Bear Ward, Great Ormond Street; and in August, he was listed for a heart transplant.

Ever resourceful and content, Sebastian spent the long days in hospital painting, gluing, and sticking stickers on anything or anybody who passed. He continued to maintain a harem of nurses and doctors and was often to be found enjoying a 'cug' (Sebastian's word for cuddle) in the arms of his devoted fans.

Always a trailblazer, Sebastian was the first child in the UK to return home with a Milrinone CADD Pump, and we were lucky to spend three precious weeks together again as a family of four.

Sebastian never got his heart. Having waited patiently for so long, he died at Great Ormond Street, in our arms, surrounded by those who loved him, aged two years, five months. Always courageous, he decided that his path led in a different direction, and we had to say farewell though it broke our hearts.

Sebastian took the hand that was dealt him, and played it beautifully. We've drawn strength from his spirit and his dignity to go forwards, to be better people, and never to forget him.



Sebastian



Sebastian and Gabriel

Spring 2014 — President's Report BSFCa

By Lynn Elwood, President, Barth Syndrome Foundation of Canada



Lynn and Adam (age 24) display BSFCa bears.
(Photo courtesy of BSFCa 2013)

This has been a year of organizational restructuring. The Canadian Government changed some of the statutory rules affecting charities, and this required that we review and update our operating by-laws. Considerable time was spent in attending government sessions, reviewing their material and working through what we thought might need to be changed in our by-laws. We enlisted help from a legal firm that specializes in charity and not-for-profit organizations, got their advice and benchmarked against other Canadian charities.

Our AGM is soon, and as usual, we will have a good balance between routine business and social interaction. The discussion and passing of our new by-laws and the opportunity for everyone to participate in the unusual activity of 'Glow in the Dark Mini Golf' will strengthen our organization in the years to come.

While the board has spent time focusing on our governance and structure, the executive and larger set of volunteers has continued to find creative ways to raise awareness and funds and to leverage their passion for the good of the organization. This year (in fact this started last year), many of our skilled craft specialists created hundreds of pairs

of mittens and scarves and dozens of dressed bears. For those joining the conference, you will see and be able to buy these treasures made with so much passion and heart. We cannot thank our volunteers enough for all the many hours they have spent in support of our efforts!

Some of the great things about our Foundation and the extended set of "Friends of Barth" we have are the surprises we get when people help in unexpected ways. We continue to have people who create and run personal fundraisers such as events and draws. In the last months we have also benefited from memorial donations on the passing of some of our Friends of Barth, including some who joined us as "Friends of Friends". It is truly inspirational when we see a notice that one of our personal friends or donors has chosen to help the organization and our affected individuals in their final wishes. Thank you to all of our Friends of Barth for your kind and thoughtful help.

This year, we once again look forward to the Barth conference, where we will spend time with our Barth family learning, laughing and sharing. The Canadian group is a big part of the conference, helping to organize and funding the attendance of Canadian researchers. We are also sponsoring the Science & Medical Poster Session, a conference breakfast and some fun at the Photo Booth. On top of that, a number of individual sponsorships by Canadians will help to defray the cost of the conference for us all. The conference is always a special week to remember in our lives, and most importantly, in the lives of our affected individuals. We are glad to be a part of it and can't wait to see everyone there!



BSFCa 2014 Planning Session

By Susan Hone, Secretary, Barth Syndrome Foundation of Canada



(L-R) Susan Hone, Lynn Elwood, Cathy Ritter, Lois Galbraith, Chris Hope and Carol Wilks. (Photo courtesy of BSFCa 2014)

The BSFCa planning weekend was held on November 22–24, 2013. Once again, Lois Galbraith and Carol Wilks volunteered their homes in the remote, wooded area of Lake Kasshabog, Ontario to hold the meeting. In attendance were Lynn Elwood, Cathy Ritter, Chris Hope, Susan Hone, Lois Galbraith, Carol Wilks and our chef/dishwasher/board advisor, Les Morris.

Friday evening, we did our customary year in review and tweaked the agenda for the rest of the weekend. Our focus for 2014 continues to be affected individuals and their families and what we can do to make living with Barth syndrome a little easier. We had a lengthy discussion about fundraising which unfortunately has to remain our number one priority if we want to be able to accomplish our mission. Other discussions included how to increase awareness of Barth syndrome, the upcoming 2014 Conference, what's new in the science and medicine area and setting the budget for 2014. We always seem to have more to discuss than the time allows.

A special thank you to Les, Lois and Carol for their hospitality and great food. Having home cooked meals and no hotel expenses keeps our administrative costs at a minimum, something BSFCa prides itself on. An added bonus this year was the addition of wireless internet at both homes; no more dial-up internet!



All photos were taken at BSFCa's AGM/Outreach 2013 and 2013 Golf Tournament.



Canadians Love their Golf, their Golfers and their Guys!

By Lois Galbraith, Volunteer



Adam, Chris, Robert and Travis
(Photo courtesy of BSFCa 2013)

September 8, 2014 will mark our TENTH annual golf tournament. These golf days have all been very exciting and filled with good fun, much laughter and great friendship.

Our plans for this year's "BSFCa Driving for the Cure" are well under way, as we have been preparing the registration brochure, making a budget and soliciting corporate sponsors. It is our hope that this year's golf day will be big, better and bouncing with more fun!

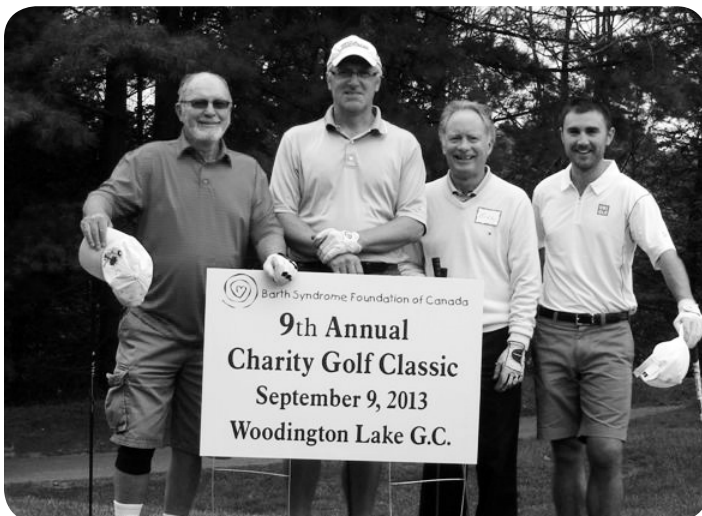
We, of course, thank our golfers and volunteers from all of our tournament days for their continued support and dedication to the BSF of Canada. To date, we have raised approximately \$165,000 for the BSFCa plans, activities, research and family support. We are proud to be fulfilling the vision of...*"Enhancing the lives and outcomes of Canadian individuals and families affected by Barth syndrome."* We still have much to do!

To succeed takes determination, organization, drive, fundraising and a strong desire to beat this disorder. We owe much of our success to many people who believe in the BSFCa and encourage and support us in so many ways.

We must commend our devoted sponsors who have been instrumental in our successes over the past ten years. The Buss Megg Society have sponsored our luncheon every year and will again this year. Thanks also to major sponsors Hope Aero, Ian Morris (Jones DesLauriers), Rick Kritschgau (Vision 2000) and Jamie Sheppard (Woodington Lake Golf Club). We also have had hole sponsors galore supporting our worthwhile cause.

Each of the ten years, we have enjoyed the company of four or five of our guys affected by Barth syndrome. They have come out to golf and to be volunteers and ambassadors on these days. They have been an inspiration to all of us. Our Saskatchewan family continues to join us for the day accompanied by various members of their family from year to year. For several years, three of our Floridian friends, Jan Kugelmann, Sharon Olson and Joanie Weaver, have made the trek to beautiful Ontario to celebrate the day with us. They certainly have added a special zest to the golf day.

Anyone who would like to join us for a day on the links, volunteer on the day, sponsor a hole, offer your talents or skills, or simply make a financial donation to the day....please contact me directly at 705.877.3159 or at lois.galbraith@sympatico.ca.



2013 Champions Ken, Rob, Bill and Tyler.
(Photo courtesy of BSFCa 2013)



Cathy, Chris, Steve and Ryan.
(Photo courtesy of BSFCa 2013)

News From Association Barth France

By Florence Mannes, Chair, Association Barth France

2013 Annual Overview

Barth France has closed its 3rd year, with EUR 81.000 raised this year. 80% of the funds was raised at different fundraising events, such as Ironman races, a golf tournament, a gospel concert, a poker tournament, a charity dinner, etc.

During this year, Barth France funded research grant programs, in Italy and in France:

1. Barth France decided to fund one of the research programs that the Barth Syndrome Foundation awarded in the 2012 grant cycle. This was the program of Dr. Angela Corcelli, from the University of Bari in Italy, to work on a potential new method for the screening of patients for Barth syndrome (BTHS). As a correct diagnosis for BTHS is often missed, and BTHS is globally underestimated, Dr. Corcelli's project would allow a quick and simple determination. Even if further experimentation is needed to validate the method, the preliminary data available show that BTHS can be easily identified. Barth France funded this work in the amount of US \$36,000.
2. Barth France belongs and contributes actively to the functioning of the French National Register for Neutropenia. This group registers each French patient diagnosed with neutropenia, with a link to any other pathology the patient may suffer from. We believe it is essential to help this registry to be kept alive and up to date, as it is an easy way to know the French Barth population dealing with this aspect of Barth syndrome.

2014 plans

Some "traditional" events...

Barth France is working hard to propose different events to donors over the course of the year:

- March: Gospel Concert (2nd year)
- April: Paris Marathon (3rd year)
- June: Heroes Race (6 km, with 20 runners for Barth France) (3rd year)
- July: Ironman (4th year)
- August: Golf Tournament (4th year)
- November: Poker Tournament (3rd year)

...and some new events...

Some other fundraising projects are being worked on as well:

- January: Charity Dinner
- May: Photo Session for Mothers' Day
- September: Garage Sale



If you would like to help Barth France organize these events, or if you have any ideas for raising funds and awareness of Barth syndrome, please do not hesitate to contact us (contact@barthfrance.com).

Research Grants

In the 2013 grant cycle, the Barth Syndrome Foundation awarded six research projects, among which is Dr. Reynolds' systematic investigation into sensory and motor-based feeding issues in boys/young men with Barth syndrome. Her previous work identified sensory issues related to feeding and eating that were ubiquitous in Barth children, with some behaviors such as strong gag reflex identifiable early in development. The purpose of this study is to build upon the preliminary findings and expand the methodologies for data collection. Barth France decided to fund this research program for US \$18,732 (see page 8 for more details).

Barth Syndrome Foundation Conference in Florida

As in 2012, Barth France will attend the upcoming Barth Syndrome Foundation Conference in June in Florida. The association will provide French families with a summary of this conference, as many families cannot attend, mainly due to the language barrier. We are very pleased that Dr. Jean Donadieu, hematologist, will be presenting during the Scientific and Medical Sessions at the Conference. Dr. Donadieu is responsible for the French National Register for Neutropenia. Our goal for the 2016 Conference would be to have more French-speaking families attending!

(Cont'd on page 26)

(Cont'd from page 25)

A French Clinic Soon

Following the visit of the Bristol Clinic Team to Paris last September, French doctors agreed to set up a French clinic in Paris in order to offer French-speaking Barth boys a one-day multidisciplinary check-up focusing on Barth syndrome. The very first clinic will be held on September 15, 2014. All of the ten French boys with Barth syndrome known to us will be invited, and we hope that many will be able to come. Barth France will organize a social event the previous day to help the boys get to know each other and to make this day special.

French Doctors Raise Awareness

In order to raise awareness of Barth syndrome in France, doctors from different specialties such as cardiology, hematology, genetics, and mitochondria have decided to publish guidelines on diagnosis and treatment for Barth syndrome. This article will facilitate early diagnosis of Barth syndrome in France.

Some Words from a Fundraiser

By Mathieu Guedon

I work for Alcoa Fastening Systems which is a division of the Aluminum producer ALCOA (Aluminum Company of America). Through their charitable foundation, ALCOA is able to support organizations like Barth France and encourage their employees in wellness activities. As a triathlete, it is an opportunity for me to combine my passion with a grant from Alcoa for Barth. I met Philippe two years ago at the running show before the Paris Marathon. I was immediately interested in helping Barth France because it supports research into a syndrome which affects children. It's the second time we have supported Barth France, and I think we will join in other events..."



Mathieu and Xavier at Eco Trail Paris (80 k)



Anne Sophie, Julien, Aurélie and Nicolas at Eco Trail Paris (30 k)

I Attended a Cardiology Conference for Association Barth France

By Laure Mercier

On March 29, on behalf of Barth France, I attended a one-day conference held by the French Association of Cardiology. The morning sessions were dedicated to the implications of cardiac diseases on everyday life: how to deal with cardiac diseases at school, and how to assure as good scholarship as possible. Another session was about dealing with stress when suffering from a cardiac disease. After a social lunch, there were five different sessions, mainly updates on research (not specifically dedicated to Barth syndrome, but to general cardiomyopathies), the new technologies for ICD, genetic testing, etc. Finally, an association came to explain how they set up a Therapeutic Education Program.

I am still working on a brief summary of the key points of this day, which Barth France could send to the French families who are in contact with the association.

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Brayden (age 4) 2013



Devin (age 9) and Henry (age 5) 2013

(Photos courtesy of BSF)

Power of Kindness

2013-2014 ~ Time, Advice and Donations



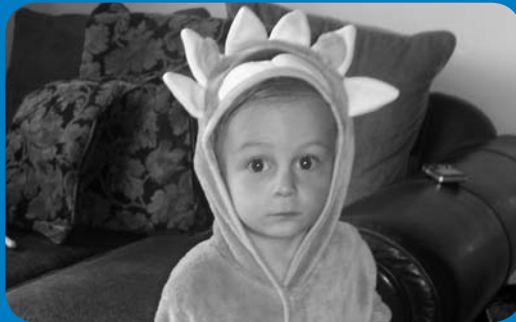
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William (age 2) goes wild ²⁰¹³



Dummer Golf Club Seniors' Captain Eddie Shaw with £1505 for BST's Annick Manton ²⁰¹⁴



A proud Mitchell (age 8) after auntie Patsy Jones' London Marathon ²⁰¹⁴

(Photos courtesy of BStrust)



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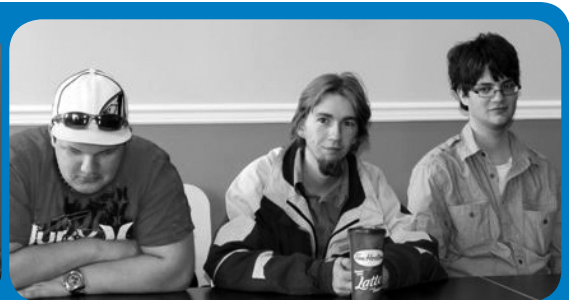
Jared, age 20



Adam, age 24



Robert, age 28



Joshua, Adam, and Ryan, age 22



Barth Syndrome Foundation

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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:

- **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