

Barth Syndrome Journal

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



2010 BSF Conference Highlights Many Advances in Science and Medicine

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

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“Given the breadth and quality of the work presented at this latest conference, the next meeting in 2012 is sure to reveal even further progress towards a specific treatment for this complex but compelling rare disease.”

The Barth Syndrome International Scientific and Medical Conference, held during the last week in July at the Renaissance at SeaWorld hotel in Orlando, Florida, was truly unique. Over 65 scientists, physicians, and healthcare professionals met to hear 26 speakers and to discuss the progress in Barth syndrome (BTHS) research and how it may lead to better treatments. In a separate but parallel set of meetings, nearly 50 BTHS individuals with their families also met to discuss issues of specific importance to their situation. In total, nearly 350 people attended this dual-track conference. The informal mixing of this diverse group of individuals at common meals, at the Poster Session and at the social function, now a traditional part of this conference series, is valuable and encouraging.

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Barth Syndrome Mouse Model is Very Promising

By Zaza Khuchua, PhD, Research Associate Professor, Molecular and Cardiovascular Biology, Children's Hospital Medical Center, Cincinnati, OH



It was spring of 2009 when I received an e-mail message from Dr. Matthew Toth, PhD, Science Director of the Barth Syndrome Foundation, asking whether I wanted to participate in the initial characterization of tafazzin-deficient mice that had been created under a BSF contract with TaconicArtemis GmbH, a European firm. At that time, I had already invested significant efforts into the generation of the animal model of human Barth syndrome (BTHS). I was very excited about this new prospect. Dr. Toth sent me the initial documentation about this project, and I was amazed by the amount of the groundwork that had already been done by various people in different institutions around the world to make tafazzin-knockdown mice available for researchers.

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Letter from Our President

BSF ~ A Tenacious Community

By Valerie (Shelley) Bowen, President, Barth Syndrome Foundation



Shelley, with her son, at BSF's 2008 conference.

It has been nearly a year since I last wrote to the readership of the Barth Syndrome Journal. I confess that was the most difficult letter I have ever had to write. I had to share the news of my son Michael's death which had occurred just days before. I was not sure what the future would hold for me. I just knew that life without Michael was the life I had spent a lifetime trying to prevent.

From the moment of Michael's death, nothing was ever to be the same. It wasn't until Michael's death that I realized the depth of love I had for all of our Barth "boys." I knew I loved them. After all, I wouldn't do what I do to save them if I didn't. I just didn't know the depth of love I had for them all. In truth, it has been the boys in our extended Barth family who have saved me. Michael's life has allowed me to find that comfort.

Over the past months I have struggled to find my center. The equilibrium of my identity had been so intertwined with Michael. I wasn't sure how I could ever find a way to do what I had done so naturally before. How would I ever be able to offer hope to a family when I had lost both of my boys to this dreadful disease called Barth syndrome? Until December 9, 2009, Michael represented the greatest means by which to offer hope to those parents who had a son just diagnosed with Barth syndrome. Even after Michael's death I found my courage by reflecting upon his life. His life was lived fully, never wasting a moment in fear of what might happen in the future.

In Michael's life each "first" milestone seemed sweeter because there had been no promise that it would ever happen at all. So, when those "firsts" did come around, they were all the more precious. I have reflected upon the events of Michael's life in recent months, knowing now that there also were "last" moments. I was blissfully unaware of the significance of these moments when they occurred, yet painfully aware of them as being the "last" after the loss of Michael.

In the months following Michael's death it was necessary to plan a conference with painful cognition that he would not be there. I was reminded of my loss as I looked at the names on the clinic schedules that I had prepared for each of the boys. Michael's name was not there. Nothing I ever did again would ever make a difference for Michael.

I pulled a notebook from my desk to jot some notes. Unbeknownst to me this was Michael's notebook from school. He was only able to attend one day of college in 2009 because he was diagnosed with heart failure the evening following his first day back. His assignment was to write an essay about an event of personal significant importance. There before me were the words of my son, written in his own handwriting, describing such an event. It was the conference he considered to be an event of significant importance. He wrote, "*The BSF conference is personally significant to me. It is meaningful to me because during that time I have the rare opportunity to be surrounded by those I care about and those who care about me. During the conference people are there who understand without words. But what is most important to me is during this event I have the opportunity to participate in research that will help others even after I'm not here anymore.*" That was all he wrote. Once again, it was Michael's life that offered me hope and clarity.

(Cont'd on page 3)

(Cont'd from page 2)

There are moments in life when one is fully aware of those “firsts” that happen. In contrast, it isn’t until later that you recognize those moments that become the “lasts.” The first moments in life offer hope and happiness. Michael’s life continues to amaze me and provide hope. Profound moments of enlightenment arise from those times in life when we appreciate the importance of being in the moment. Those unpredictable moments come without warning. We don’t know how, when, why, or where they will happen. We just know it is important to pay attention when they happen, as they provide the memories we will recall most fondly in the future. In the blink of an eye, the present becomes the past. During the 2010 conference, I experienced many such moments. I could never have anticipated how these boys, many now young men that I have spent a lifetime trying to keep safe from harm, were stepping in to save me.

In my last letter to you, I shared Michael’s essay, *Fear Is Not My Option*. In that essay he wrote, *“I will not waste my life by living in fear. I have made the choice not to live in fear but moreover to be fearless. Fear paralyzes where courage sets us free. I will live my life to the fullest. I will not consider what will come of me. I will come and I will go. That has been defined for me. If I am fortunate enough to draw my first breath there will be a last breath that concludes my life. It is out of my power to define when those breaths occur. I can make the most of each breath in between....”*. There will be “lasts,” so why waste the present looking for something that we can’t define? Why waste the present in regretting the fact that we didn’t recognize those moments as the “lasts?” Comfort comes when we embrace the fact that those moments occurred at all.

I want more. My son didn’t make compromises when it came to living. He didn’t have it easy, but he never used that as an excuse for not making the most of his life. During the conference in July, I saw that same tenacity in each of our boys. They say thank you by living their lives to the fullest. What greater thank you could anyone offer?

I want all of these boys to experience happy “firsts.” I want no one else to experience painful “lasts.” I want everyone to do what you can now to make sure this happens. Our boys deserve it, and they need your help. They will do the rest!



Candids taken at BSF’s 2010 conference. (Photos courtesy of Cherie Schrader.)

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2010 BSF Conference Highlights Many Advances in Science and Medicine

(Cont'd from page 1)

This was the fifth international conference hosted by the Barth Syndrome Foundation (BSF). Conferences are held every two years, and 2010 marks the 10th anniversary of the founding of BSF by family members of BTHS individuals. Many of the speakers over the two days of the Scientific/Medical (Sci/Med) sessions of the conference were BSF Research Grant recipients. The chairpersons for the Sci/Med sessions were: Richard I. Kelley, MD, PhD (Johns Hopkins University, Baltimore, MD); Michael Schlame, MD (New York University, New York, NY); Barry J. Byrne, MD, PhD (University of Florida, Gainesville, FL); and Miriam Greenberg, PhD (Wayne State University, Detroit, MI).

Also included this year was a keynote lecture, “**The Pathophysiology of Mitochondrial Disease,**” which was delivered by Professor Douglas C. Wallace, PhD, Director of the Center of Mitochondrial and Epigenomic Medicine, Children’s Hospital of Philadelphia and University of Pennsylvania. The Science/Medicine sessions of the 2010 conference were funded in part by grants from the Office of Rare Diseases Research and the National Heart, Lung and Blood Institute of the National Institutes of Health.



Dr. Michael Kiebish presents on shRNA tafazzin knockdown model of BTHS.

Animal Models of Barth Syndrome

Animal models of a human disease are often a valuable tool in finding and developing clinical treatments, and leading off the Sci/Med sessions were the first reports of the mouse model of BTHS developed by BSF. These initial reports of the BTHS mouse were quite encouraging. Zaza Khuchua, PhD (Cincinnati Children’s Hospital Medical Center, Cincinnati, OH), and Michael A. Kiebish, PhD (Washington University School of Medicine, St. Louis, MO) revealed that this “knockdown” mouse model possesses the expected biochemical abnormalities and also eventually develops the heart problems reminiscent of BTHS.

Other animal models, including fruit flies (Mindong Ren, PhD, New York University, New York, NY) and rat models of heart failure (Genevieve Sparagna, PhD, University of Colorado, Boulder, CO), continue to add to our understanding of BTHS and provide new ways to think about potential treatments. BSF intends to distribute these BTHS mice to any investigator, and we expect a substantial increase in the number of new BTHS researchers since they will now have a powerful model system in which to test their hypotheses. Currently these animals are being studied in five laboratories in the US and Europe.



Dr. Michael Schlame leads the brainstorming about future directions in BTHS research.

Barth Syndrome Pathophysiology

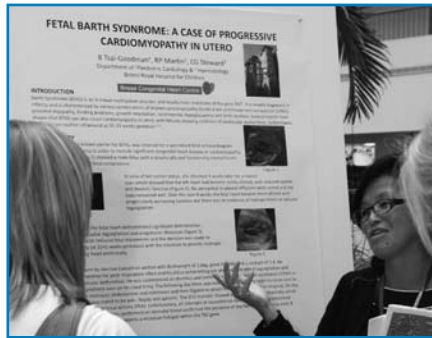
BTHS is considered a unique mitochondrial disease. Mitochondria are those distinct parts of the cell which provide it with energy; defective mitochondria have been implicated as the cause of several human diseases and as one of the main causes of the natural aging process. Using exercise as therapeutic treatment, Mark Tarnopolsky, MD, PhD (McMaster University, Hamilton, Ontario) showed that mitochondrial DNA deletions in elderly people can be reversed by exercise. Dr. Tarnopolsky then discussed what this may imply for BTHS. In one of the first clinical research projects funded in part by the BSF Research Grant Program, Todd Cade, PT, PhD (Washington University School of Medicine, St. Louis, MO) is now testing whether supervised aerobic exercise training (cardiac rehabilitation) is beneficial for BTHS individuals.



Some of the dedicated doctors (and staff) involved in the cardiology clinic at BSF’s 2010 conference.

Dr. Cade, along with Carolyn Spencer, MD and Amy Roberts, MD (both at Children’s Hospital Boston, Boston, MA) also presented the unique physiological characteristics of BTHS individuals. These physiological data, much of which have been collected at the conference “clinic,” provide us with a better understanding of this disorder. The Barth Syndrome Medical Database & BioRepository (BRR), which is supported by BSF and now also by the Children’s Hospital Boston, will collect and store these data, other relevant medical information and numerous biological samples for researchers to use.

(Cont'd on page 5)



Dr. Tsai-Goodman at Poster Session.

Of particular note for the BSF community was a presentation by Colin Steward, FRCP, FRCPCH, PhD (Bristol Royal Hospital for Children, England). Dr. Steward has found many previously unrecognized cases of BTHS in the Bristol area of England by pursuing the neutropenia aspect of this disease and by doggedly investigating unexplained male fetal deaths in several affected families (Barth syndrome: an X-linked cause of fetal cardiomyopathy and stillbirth. See page 15 for complete journal citation.) Dr. Steward, along with the assistance of Michaela Damin and the Barth Syndrome Trust (the UK affiliate of BSF), has set up a National Specialized Service for BTHS within the National Health Service in the UK, and he provided insights for establishing a similar group in the US.

Mitochondrial Dysfunction and Its Impact on Human Disease Lipids, Tafazzin, and Mitochondrial Metabolism in Barth Syndrome

Human diseases that resemble BTHS also have a lot to teach us. Charles Hoppel, MD (Case Western Reserve University, Cleveland, OH) provided an overview of mitochondrial diseases by focusing on oxidative phosphorylation defects. Robert E. Jensen, PhD (Johns Hopkins University, Baltimore, MD) illuminated the important parallels between BTHS and Dilated Cardiomyopathy with Ataxia (DCMA) and discussed how mitochondrial protein transport may link the common symptoms of these two genetically distinct but similar, rare conditions. By relating what we know about other human diseases that share characteristics with BTHS, we may be able to formulate treatment ideas for both BTHS and for these other diseases.

The number and breadth of the Science/Medicine presentations show that BTHS research has come of age. These sessions featured presentations ranging from lipid biochemistry; the composition, intermediary metabolism, and dynamic movements of mitochondria; and the screening for genes/compounds that reverse the genetic defects of the BTHS mutation. Most of these presentations were made possible, in part, by funding provided by BSF and our affiliates through its Research Grant Program. The eight years of this grant program, along with the conference “clinic,” the BRR, and the dedicated efforts/publications of BTHS researchers and BSF members have provided the scientific/clinical foundation to guide investigators to make the progress that will translate into real clinical benefits.

In addition to the Science/Medicine presentations, 12 posters were presented in a separate session that was well received by both the Sci/Med attendees and by the families of BTHS individuals. The interactions between these two groups are extremely important as both groups get to know and appreciate the details and the problems each face—a perspective that often is lacking in other science/medicine-oriented meetings.



John Wilkins and Jessica Wiederspan present the 2010 Varner Award to Dr. Toniolo.

Varner Award for Pioneers in Science and Medicine

The Varner Award for Pioneers in Science and Medicine was awarded to Daniela Toniolo, PhD, and posthumously to Peter Vreken, PhD. Dr. Toniolo (Research Director, National Research Council of Italy, DIBIT-San Raffaele Research Institute, Milano, Italy) was recognized for her discovery of the tafazzin gene in 1996, and the late Dr. Vreken (Academic Medical Center, University of Amsterdam, Amsterdam, The Netherlands) was recognized as the first to publish on the cardiolipin abnormalities of BTHS individuals in 2000. It was particularly appropriate to mark these significant scientific milestones at BSF's 10th anniversary celebration.

Barth Syndrome Clinic

Also unique to this conference series is the information-gathering session or “clinic” that is provided by BTHS physicians, healthcare workers and researchers. This “clinic” serves at least two purposes: (1) it allows the efficient collection of physiological data and historical medical information from patients with this rare disease; and (2) it provides opportunities for patients and patient family members to meet with physicians who have a substantial experience in treating BTHS individuals. In 2010, six distinct IRB-approved protocols were participated in by many of the BTHS individuals who attended the “clinic.” Most of the data collected are expected to lead to publications and/or ultimately to be available through the BRR which is open to all interested researchers.

In Summary

The 2010 Conference included the greatest number of speakers in its history, only some of whom could be mentioned here due to space considerations. The Science/Medicine sessions were packed with new information and new developments. All of the individual presentations, for both the Sci/Med sessions and the Family sessions, were recorded on DVDs and will soon be available for a nominal cost by contacting BSF (www.barthsyndrome.org). Given the breadth and quality of the work presented at this latest conference, the next meeting in 2012 is sure to reveal even further progress towards a specific treatment for this complex but compelling rare disease.

2010 Conference Attendee Testimonials

“Having never attended a Barth syndrome conference before, I could never have anticipated the sense of community and urgency that encompasses the Barth Syndrome Foundation’s families and dedicated scientists. In my opinion, no conference exists that integrates and recapitulates the emotion and purpose of synergistically merging the efforts of the research community with that of the passionate goal of the families toward developing a therapeutic strategy to treat young boys/men affected with this disease. You might walk into the conference as a scientist, but as I found, you walk out of the conference as an adopted family member working for the same cause and battling for the same goal. ...” ~ Michael Kiebish, PhD, Xianlin Han Laboratory, Washington University School of Medicine, St. Louis, MO

“A huge thank you to all the volunteers who made this conference a spectacular event. I wish I could explain to all ya’ll (LOL) how much it meant to our family, how much it touched us, how much it taught us, and how grateful we are having met everyone. When our son’s doctors first told us that our son had a rare disorder called Barth syndrome, I was very confused, angry and frightened. I reached out to this community and was immediately welcomed with open arms. I was given wonderful advice and many promises that in this community WE DO NOT GIVE UP on OUR BOYS! How true I have found this to be.” ~ Brie, BTHS Family, Kentucky, USA

“It was my first time at an International Meeting of the Barth Syndrome Foundation, 14 years after identification of the gene, and it was a very exciting experience. I had followed the work done by the Foundation through the BS Journal that I received regularly, but the active participation in the meeting of so many scientists and of many patients and family members was very impressive. I think the scientific results presented at the meeting and the general atmosphere were unique, and they represented the best reward for our work. Thanks again for inviting me and giving me the opportunity to see how sometimes the results of a scientific achievement can go beyond a good publication.” ~ Daniela Toniolo, PhD, Division of Genetics & Cell Biology, San Raffaele Scientific Institute, Milano, Italy

“I am writing this on our drive home, with the bittersweet feeling of the extraordinary week we spent together laughing, crying, supporting and inspiring each other. I am going home with a “high” unmatched by any other experience I’ve had. There is so much that we need to process and reflect upon from this past week; from the informative lectures, the private conversations and life-long bonds that strengthen with each conference. We want to genuinely thank ALL who made this week a memorable one. It may be hard to envision 2012 now, but I genuinely can’t wait to reunite again with our Barth family.” ~ Amer, BTHS Family, Florida, USA

“I was very pleased to attend the 5th BSF meeting to talk about my research on X-chromosome inactivation in female carriers of Barth syndrome. I have been a clinical geneticist for almost 40 years but previously only met one family with boys with Barth syndrome. Therefore, I greatly enjoyed meeting all the boys and their families and hearing their stories, which they presented so bravely to the audience. I was also very impressed by the scientific quality of the meeting. The conference was an outstanding example of how efforts initiated by parents can contribute to increased knowledge about this rare and puzzling disorder.” ~ Karen Helene Orstavik, Department of Medical Genetics, Oslo University Hospital, Oslo, Norway

(Cont’d on page 9)

Group photo of BTHS families together with scientific and medical community at BSF’s 2010 conference. (Photo courtesy of Amanda Clark)



A Decade of Community and Education

BSF Hosts 5th International Barth Syndrome Family Conference

By Leslie Buddemeyer, Conference Committee, Barth Syndrome Foundation

When asked to write about the Family sessions at BSF's 2010 Conference, I didn't know quite where to begin. Should I describe the Family sessions, give my opinion, or go with my gut? I decided to go with my gut. My first conference was in 2004. My son had just been diagnosed with Barth syndrome (BTHS) a couple months earlier, at the age of 14 years 11 months, and, to be honest, I didn't want to attend the conference. I was too afraid of what it was going to be like for my son and my family (still just trying to grasp the diagnosis). But, I must say, I was completely blown away with the experience we had. The Family sessions, in particular, were more than I ever could have expected. And now, after having attended my fourth conference, I still feel the same way.



Families review session topic and gather questions to be addressed by session presenter.

I was inspired after attending the first conference, along with the persistence of Shelley Bowen, to become more involved. Now that I have, I have a fantastic appreciation for those who are behind the scenes putting this entire conference together. I can't begin to describe the process, but I can tell you that work on the next conference begins the day one conference ends. The topics that are to be discussed are never taken lightly. The Family sessions have to be informative and relatable—not only to returning families but to first-time attendees (many newly diagnosed) as well.

To me, the Family sessions at BSF's 2010 conference exceeded this and more. There were nearly 50 affected families in attendance, 12 of whom were first-time conference attendees. Many had travelled great distances to meet with world-renowned experts in the field of BTHS to gain the most up-to-date information about treatments and research into this disorder.



A Family session at BSF's 2010 conference.

Personally, I have never felt more connected with the physicians, scientists, specialists, and families than I did this year, and my understanding of BTHS increased as well. This was achieved beginning with the first Family session, *Cardiac Aspects of Barth Syndrome*, and continued until the last. It is my opinion that all of the speakers/presenters were very engaging with their topics. It was understandable to me, which is huge to this simple mind. I also must say that I really enjoyed having our speakers introduced by BTHS family members. Even though our stories are similar, they are each unique and compelling.

The challenging task of planning the Family sessions is trying to fit everything in. And, we always seem to want more—more time to ask questions, more time for answers, more time for discussions, and more topics. The remarkable thing is that we manage to do this. Maybe not during the actual sessions, but in the lobby, at dinner, and when we get home and log onto BSF's Family listserv—we talk. I don't know that there will ever be enough time during the actual sessions to achieve all of this, but we are connected, and every question, comment and discussion is always welcomed.



Review of session topic and gathering of questions to be addressed by presenter.

One of the most poignant moments for me was our break-out session titled *Parent Perspectives*, and I am speaking as a mom here. I was sitting there in this small room with all these mothers from around the world, most of whom I had never met before, laughing, crying, and bonding. We expressed differences, similarities, opinions, and support. This was not just about our sons—it was about our lives, as simple and as complex as they are. I came away from that experience feeling truly understood and supported. There is nothing this BSF FAMILY cannot do!

I look forward to attending the next conference, the Family sessions, and most of all seeing this (OUR) Family again!



Embracing a Rare Disease ... Becoming More Involved

By John, Conference Planning Committee, Barth Syndrome Foundation



John shares a special moment with Milosh. (Photo courtesy of Cherie Schrader.)

I was pleased to be asked to serve on the Management Committee for the 2010 Barth Syndrome Foundation's 5th International Scientific, Medical and Family Conference. I learned firsthand what a huge undertaking this wonderful event is. We started planning in November 2009, and each of the three committees I served on met at least once a month over the phone. Many details were discussed and debated, including planning how meals would be served, meeting room arrangements, what speakers would present, tee-shirt designs, and hundreds of other details. The most difficult part was planning sessions for our youth, both those affected with Barth and siblings, trying to strike a balance between having fun and imparting some knowledge about Barth syndrome. Overall, it was a very satisfying experience and great to see all the hard work pay off at the end of our week together!

The conference itself was both exhilarating and exhausting all at the same time. By far the best thing about the conference was the people and the fellowship. We are very lucky to have such an awesome group! It was great to have a chance to reconnect with old friends that we haven't seen in many years and make new friends, as well. There were twelve families who were first-time conference attendees; four of whom were newly diagnosed.

The conference was a very busy time for all of us. Breakfast was at 8 AM, and most nights we would stay out by the pool swimming and talking until 10 PM or later. The clinics went very smoothly, thanks to our awesome volunteers who kept doctors, scientists and families on schedule. Our sessions were very informative and covered topics ranging from exercise, independence and financial issues to cardiac concerns. There was also a chance for the younger guys with Barth syndrome to ask questions of the older guys with Barth syndrome, which was a great experience for both groups.

Overall I found the conference to be a wonderful experience.



Some of our BTHS youth, young men, and siblings at BSF's 2010 conference. (Photo courtesy of Amanda Clark.)

Barth Syndrome Clinical Trial Included on a Major Website

Barth syndrome (BTHS) clinical research has made it to the "big time," as a current BTHS study ([Exercise Training in Barth Syndrome](#); Principal Investigator: William T. Cade, PT, PhD, Washington University School of Medicine) is now included on [ClinicalTrials.gov](#)—the most prominent website for such projects. Dr. Cade and his colleagues are applying the discoveries that they have made at the BSF conferences to ask important medical and quality-of-life questions. They are investigating whether a certain therapy, namely aerobic exercise or cardiac rehabilitation exercises, will be helpful for BTHS individuals. This type of clinical trial is referred to as a pilot/feasibility/proof-of-concept trial and follows from the investigators' observations that severe exercise intolerance in adolescents with BTHS is mediated by impaired skeletal muscle oxygen extraction and utilization. The goal of this study is to collect preliminary data on the following hypothesis: Supervised aerobic exercise training will improve skeletal muscle oxygen extraction/utilization, left ventricular function, peak exercise tolerance, cardio-autonomic function and quality of life, and will be found safe in adolescents and young adults with BTHS.

For more information, please visit <http://clinicaltrials.gov/ct2/show/NCT01194141>.

2010 Conference Attendee Testimonials

“Our family made the trip from Scotland to Orlando to attend the 2010 conference because it is the one place where other people “get” what Barth syndrome is and what it means to your son and family. You can completely and utterly immerse your full family in this wonderful group of people. We always find that we can learn so much from the other families that attend, and the time spent at the clinics and family sessions are just as important as time spent around the pool with the other families sharing experiences, love, information and supporting one and other. If your family has not attended one of these conferences then I would urge you to try to, as you cannot buy the fountain of information that all the families are willing to share with you and your family. It truly is a remarkable group to be part of, and my family is very grateful to each and every person who works so tirelessly for the benefit of us all.” ~ Allanna, *BTHS Family, Scotland*

“I have been involved with the Barth boys now for over six years, and I am still learning. There are so many questions, and the answers are coming through but slowly. I really enjoyed the Barth Syndrome Foundation Conference in Florida, as it gave me the opportunity to meet other people who are also interested in helping these boys, from scientists to medical doctors to parents, friends and family! I have never come across any medical meeting where scientists, medical doctors and patients sit next to each other and learn from each other. A real gem of an opportunity!” ~ Beverly Tsai-Goodman, *BM, MD, FRCP, PG Cert Med Ed, Department of Paediatric Cardiology, Bristol Royal Hospital for Children, Bristol, England*

“I don’t think there are adequate words to convey what we all got out of the conference this year. In one week, we made friendships and established bonds that people struggle to make in a lifetime. The closeness of this group is both welcoming and enveloping, and you leave feeling like you have known everyone for a lifetime. We gained information from the wonderful doctors, and we were able to ask questions we struggled to get answers to at home. Most of all, our son felt at home. He felt he was where he belonged, where he was loved and not judged; he said “Mum at the conference I was normal,” and to him that meant everything. We can’t wait for 2012 when we can do it all again. ~ Donna, *BTHS Family, Australia*

“I would sincerely like to thank BSF for inviting me to speak at the 2010 meeting in Orlando. This was my first BSF meeting and it was a very enlightening academic and scientific experience since I had very little exposure to Barth syndrome and the underlying pathology prior to attending this conference. More importantly, although I have attended numerous scientific meetings throughout my career, I have never had the opportunity to interact with family members and individuals with a particular disease affliction. This interaction had a profound impact on me, and certainly put all of the basic science that we do in the laboratory into perspective. I hope to have the opportunity to be involved with this conference and BSF in years to come! ~ Peter Adhietty, *PhD, Department of Applied Physiology and Kinesiology, University of Florida, Gainesville, Florida, USA*

“It was a great pleasure to meet long known, never personally seen friends. And it was very valuable to talk to the world’s leading scientists and physicians of Barth syndrome.” ~ Sonja, *BTHS Family, Germany*

“The BSF meeting in Orlando this past July was like no other meeting that I have ever been to before: informative, cutting edge, inspiring, international, fun, productive, unforgettable and historical. Having families, boys and men with Barth syndrome, scientists and clinicians all in the same place was exhilarating and overwhelming. So much hope is born from the togetherness and the science. Simultaneously, everyone embraces the struggle of a life-changing illness. I know of no other association or group who does it the Barth way.” ~ John Saroyan, *MD, FAAP, Departments of Anesthesiology and Pediatrics, Columbia University College of Physicians and Surgeons, New York, New York, USA*

“...This conference was so full of hope. We met boys, parents, doctors, family members, all of them dedicated to one single goal: helping these boys, to which the sentence of Gandhi applies so well: “Strength does not come from physical capacity. It comes from an indomitable will. ...” ~ Florence, *BTHS Family, France*



Group photo of Barth boys, young men and siblings at BSF's 2010 conference. (Photo courtesy of Amanda Clark)

Barth Syndrome Mouse Model is Very Promising

(Cont'd from page 1)

“I firmly believe that the mouse model of BTHS and the collective effort of researchers will result in applicable therapeutic strategies to treat Barth syndrome patients and, perhaps, mitochondrial dysfunction in general.”

In June of 2009, we received two male transgenic mice from Germany and placed them in quarantine for three weeks. Our immediate task was to establish a breeding colony and ensure the safety of this valuable mouse strain. We were relieved when the mice produced the first offspring.

After the breeding colony had been established, the mice were distributed

among researchers from different institutions. In 2009, we received requests for tafazzin-knockdown mice from at least five investigators from the US and Canada.

At that time, nothing was known about these mice. We all were very curious to see whether and to what degree tafazzin-knockdown mice recapitulate symptoms of human BTHS patients. To initiate tafazzin-knockdown, the mice had to be placed on a special diet containing a small amount of the drug “doxycycline” (a soluble analog of the antibiotic tetracycline). The introduction of doxycycline into the system triggers the expression of the “transgene.” Transgene is an artificial DNA construct that is inserted in the mouse genome. Once activated by doxycycline, the transgene produces small, hairpin-like RNA molecules (shRNA), which can specifically interfere and “silence” the mouse tafazzin gene.

After two months of dox-feeding, we sacrificed one mouse and confirmed the tafazzin knockdown by a special method which allows the quantitative assessment amount of any gene product in tissue. In scientific literature, this method is referred to as QRT-PCR, which stands for Quantitative Real-Time Polymerase Chain Reaction. QRT-PCR profiling showed an almost 90% reduction of tafazzin gene product in the heart, skeletal muscle and liver of these transgenic mice. This was a very important milestone in our studies, showing that tafazzin knockdown in mice indeed worked.

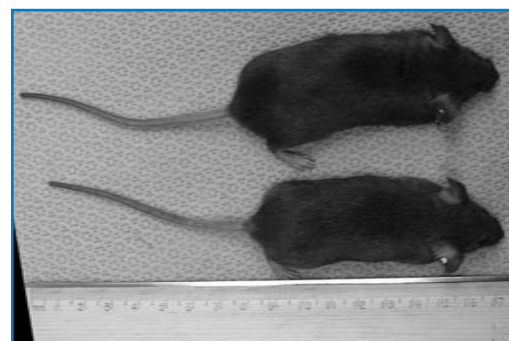
The only well-established biochemical consequence of tafazzin mutations in humans is cardiolipin deficiency. Therefore, the next question we faced was “Does tafazzin gene silencing result in cardiolipin deficiency in mice as well?” I sent frozen samples of heart, muscle and liver from tafazzin-knockdown mice, along with those from normal mice, to Dr. Frédéric Vaz at the University of Amsterdam in the Netherlands. In a few weeks, Dr. Vaz sent me back a large amount of data with calculations and graphs, showing a dramatic reduction of

tetralinoleoyl cardiolipin (L4 cardiolipin) and an accumulation of abnormal cardiolipins in all analyzed organs. These were the results we all hoped to get—cardiolipin deficiency in tafazzin-knockdown mice. We finally had a mouse model for human BTHS.

Tafazzin-knockdown mice were sent to Dr. Devrim Acehan at New York University. He acquired more than 600 images of different tissues using electron microscopy. These pictures clearly showed mitochondrial deterioration in cardiac and skeletal muscles, very similar to what we see in human cells.

The staff of the animal facility at Cincinnati Children’s Hospital Medical Center and lab personnel were instructed to observe and report any signs of developing sickness in tafazzin-knockdown mice. In fact, we hoped that we would see some sign of sickness, or as researchers call it, “phenotype,” which is an observable characteristic resulting from gene manipulation. To our surprise, the knockdown mice didn’t show any signs of sickness until the age of 8 months, which roughly corresponds to 25-30 human years. At 8 months of age, however, tafazzin-knockdown mice developed dilated cardiomyopathy and signs of heart failure. Dr. Jeanne James at Cincinnati Children’s Hospital Medical Center performed echocardiography on tafazzin-deficient mice.

We are continuing our studies of tafazzin-knockdown mice. I am in constant contact with other researchers in the BTHS community in order to coordinate our studies. I firmly believe that the mouse model of BTHS and the collective effort of researchers will result in applicable therapeutic strategies to treat BTHS patients and, perhaps, mitochondrial dysfunction in general. Because BTHS is likely to be the prototype of other mitochondrial phospholipid disorders, these studies will probably be relevant to other human disorders.



Tafazzin-deficient mouse (bottom) is a little smaller and weighs less than a normal mouse.

Newly Diagnosed Family — Henry

By Tiffini, Mother of Newly Diagnosed Son, Indianapolis, IN



Henry (center) enjoys a trip to Wrigley Field with mom and dad, Tiffini and Kevin.

Henry Michael was born on June 30, 2008, just in time to watch his first Chicago Cubs game. Unfortunately, Henry missed the game. His sugar levels were low, warranting a night in the care unit where his nurses could keep a closer eye on him.

Fortunately, the stay in the care unit led to an early diagnosis of dilated cardiomyopathy. Henry was three days old when an echocardiogram revealed that his heart was severely dilated and his ejection fraction was merely 10-15%. That day he was whisked off to the NICU, and after only nine days, his ejection fraction was at 25-30%. He was doing so great that his team of doctors released him to go home to sleep in his cozy crib.

After a couple of months out of the hospital, Henry's cardiologist suggested we seek a second opinion. After a review of Henry's echos, he was diagnosed with left ventricular noncompaction of the heart, and it was suggested that a muscle

biopsy be performed to rule out a mitochondrial disorder, just in case Henry would need to be placed on the heart transplant waiting list. That was shocking to hear, and we needed to educate ourselves. After a little on-line research, we suspected Henry had Barth syndrome (BTSH). We mentioned this to one of Henry's former doctors who dismissed the idea. While we lack medical degrees and have the utmost respect for all those who have cared for Henry, we wish that particular doctor hadn't been so dismissive, and, in hindsight, we should have insisted on a genetic test.

Barth syndrome is such a rare disorder, and we just couldn't believe Henry could have it. Although we also had that moment of thinking, "someone has to have it, so why not Henry?" Henry was four months old when his muscle biopsy came back positive for abnormal mitochondria, which made BTSH even more realistic. He had many of the symptoms: left ventricular noncompaction, abnormal mitochondria, and he was below the 3rd percentile in height and weight.

In February 2010, Henry was admitted to Riley Hospital for Children due to a viral infection. His neutrophil count was zero, making any bacterial infection very dangerous. The hematologist said that he thought Henry had something called "neutropenia." We said, "if he has neutropenia then he must have BTSH." The hematologist said they'd draw blood immediately and send it off to the diagnostic laboratory at Baylor College of Medicine for genetic testing. We'd read about neutropenia during our research, but we learned what it really meant after watching a segment on the TODAY Show that featured the Barth Syndrome Foundation (BSF).

As we suspected, on March 10, 2010, Henry's genetic test came back positive for BTSH. We were grateful that we finally had an answer. A weight had been lifted from our shoulders, and although we felt very educated about BTSH through the research we had done, especially from the information gained from BSF's website, we couldn't wait to learn more. We contacted BSF immediately after receiving the results. If we could help in any way with the research to find a treatment or a cure we wanted to get started straight away. One boy could have all of the answers, and maybe that one boy was Henry.

Shelley Bowen, President of the Foundation, called us that evening, and we talked for hours. She told us about the 2010 International Conference, and we said that we'd be there. The conference was phenomenal! Researchers, doctors and families from all over the world gathered in Florida to help find treatments and a possible cure. It was amazing to us that so many doctors and researchers were interested in a disorder that only affects approximately 150 families worldwide (although BTSH is believed to be under-diagnosed by many physicians).

The information we came home with is indispensable. We realized that the research the scientists are doing is not only helping our boys with BTSH, but it is also helping kids and adults with other diseases such as cancer, diabetes and mitochondrial disorders. We knew the researchers and doctors were not just people that we would meet once and never hear from again. They are now our friends. We are also very fortunate to have a fantastic team of doctors at home that are more than willing to work with the doctors and researchers we met at the conference which adds a lot of comfort to our lives. Henry could not be in better hands!

We created invaluable friendships with the other families attending the conference that will last a lifetime. We've realized that if Henry didn't have BTSH, our life would lack the richness these relationships have brought! On behalf of Henry and our entire family, we thank everyone who has become a part of our Barth syndrome extended family.

A Decade of BSF's Extraordinary Contributors

By Stephen McCurdy, Chairman, Barth Syndrome Foundation

As any child will tell you, becoming 10 years old is a big deal! At 10 you are no longer a “baby.” At 10, people can begin to see what kind of a person you might become, and if your early years were anything like mine, 10 is also a time for those who love you to reflect – sometimes with astonishment – on how you ever got this far!



Joe William (age 2) at BSF's 2010 conference. (Photo courtesy of Amanda Clark.)

Like a child, BSF started with a future full of hope and opportunity, none of which could be realized but for the unselfish contributions of a faithful family of supporters. Now, after a decade, we can begin to see the shape of a healthy, growing community that is already having a major impact on the lives of the children and families affected by Barth syndrome. Throughout this Journal are article after article chronicling the latest achievements of our community in science, medicine and family support—each moving us closer to our goals of finding effective treatments and a cure for Barth syndrome and in the meantime, caring for the families struggling to look after their affected children. Nurturing all this growth, enabling all this progress, and insuring that we have a future like the one we can only dream of today is a quiet group of supporters who have had faith in us all along; who, like proud parents, watch our progress, providing guidance and support when it is needed, but who know that a sound foundation requires both deep roots to grow as well as strong wings to fly. Our future will be what we make of it, but we cannot do it alone.

Today, we take the time to celebrate the loyal supporters who have been with us faithfully since our beginning, 10 years ago. We acknowledge with deep appreciation those whose extraordinary gifts have inspired us to achieve accomplishments that at first seemed only dreams. We shine a spotlight on those who have had the courage to ask their friends and family for support and by so doing, have enabled this community to grow to what it is today. We would not be who we are without you. Thank you, from all of us!

Our Loyal Sustainers

Out of thousands of donors, a few have been with us unflinchingly from the start. Their presence on this list is not about the size of their donations but the depth of their loyalty. Their names have been listed on our “Power of Kindness” pages in every Barth Syndrome Newsletter ever published, and we celebrate their fidelity and constancy. Like our parents, these supporters have been with us since our birth.

Our Constant Supporters		
Peter Allman	Ned & Liz Hazen	Suze & Peter Osnos
Tom Allman	Norman & Debbie Hedgcock	Rob Otte
American Express	Dr. Bruce Henricks	Aldo & Sandra Papone
Rev. Frank Bancroft	Dr. Greg Hurlbut	Bob & Jackie Purcell
Rick & Carla Berry	Barbara Kenyon	Bob & Liz Sauer
Dr. Dave Bingham	Dr. Fred Kiechel	Ted Sonderegger
Nick & Sally Bogert	Steve & Jan Kugelmann	Dr. Gene Stohs
Shelley & Michael Bowen	Dr. Martin Massengale	Dennis Stuckey
Russell Brehm	Steve & Kate McCurdy	Judy Varner
Andy & Kathy Brenner	Bill & Ginny McCurdy	Tom Varner
Charles Burmeister	Gates Minnick	Al & Elaine Viebranz
Robert & Ann Carroll	Steve & Beth Mixer	Joanne Wilkins
Dr. John Cheatham	Marc & Louise Morgenstern	Dr. Mike & Sue Wilkins
Peter & Gretchen Crowley	Bob & Diane Motzkin	
Dana Hart	Dr. Tom Osborne	

(Cont'd on page 13)

(Cont'd from page 12)

Not surprisingly, these include many of our founders and their neighbors, friends, and family. Susan Osnos is now on our Board. Aldo Papone provided sound advice before we were even incorporated. It is also striking how many doctors are included in this list like Dr. John Cheatham, a pediatric cardiologist who treated a BTHS boy (who is now an adult) for many years. The list even includes a corporate patron through gift matching and small employee grants.

Our Quiet Benefactors

These people and institutions have made extraordinary contributions to BSF over the last 10 years. It is true and fair to say that without their exceptional beneficence, we would not have a research program, the Barth Syndrome Medical Database and BioRepository, nor our biennial conference. It is also fair to say that they all hold us to very high standards. They continue to have faith in BSF because we prove every year that we are good stewards of their investments. Paula Varner provided our first and founding donation. Mrs. Annenberg gave us the initial donation that funded our Research Grant program and then continued to fund BSF for the rest of her life. Peter and Isabel Malkin's annual support helps BSF enjoy a base level of financial stability which in turn enables us to initiate multi-year programs. Our affiliates, BSTrust in the UK and BSF of Canada provide support for our shared programs, and the National Institutes of Health has provided grants to both BSF and a number of our researchers. Several of these donors, like Dr. Fung and Anis Sattar, support BSF because of our Team Will Ironmen, and the United Space Alliance is a regular supporter of the Kugelmann's golf outing. All of these friends of BSF were first friends of a BSF family who introduced them to our challenges, hopes and dreams. Today, we extend to each a warm embrace as special friends of BSF.

Our Largest Donors		
The Annenberg Foundation	The National Institutes of Health	Marc & Tracy Sernel
Dr. Paul & Allene Russell	Dr. Richard & Allene Pierson	Clarke & Patricia Bailey
Peter & Isabel Malkin	Jonathan Ledecy	Christopher & Abby McKown
Andrew Pietrini - The Lebensfeld Foundation	Paula Varner	Ann Geary
Steve & Kate McCurdy	Richard & Cynthia Blumenthal	Marilyn Lummis
Scott & Laura Malkin	Suzanne and Walter Scott Foundation	Natan & Beth Vaisman
Christina Hixson - Lied Foundation Trust	Dr. William K. Fung	Dan & Susan Semrad
Ed Pace - Lake City International Trucks, Inc.	Dillon Foundation	Samuel & Ann Mencoff
Barth Syndrome Trust	Robert Daugherty	Rusty & Jane Lewis
Mr. & Mrs. Harold Anderson	Janus Foundation	Holman Enterprises
The Frill Foundation	Seth & Amy Barad	Anis Sattar - Fergasam Garment Ind. Ltd.
American Express Foundation	Dr. Mike & Sue Wilkins	Michael & Gail Yanney
William & Dossy Lummis	Susan & Peter Osnos	United Space Alliance
Rick & Janet Sherlund	John & Sara Lindsey	Richard & Virginia Voell
Tom & Carrie Cusack	Tony L. & Debbie Campbell	BSF Canada

BSF Ambassadors

At the Barth Syndrome Conference last July, four people were acknowledged as examples of Barth ambassadors for their continuing extraordinary efforts to promote awareness and support for BSF: The Higgins family for their annual fund raising bowl-a-thon and tireless support for BSF; Cherie Schrader for the courage to use her Chicago Marathon runs to honor the memory of her nephew, Lattigo Cook, and helping to transform the pain of loss into something good and lasting; Dr. Michael Schlame for his uncanny ability to find new researchers for Barth syndrome as well as his personal financial support; and Gary Rodbell for creating *Team Will* and forming an iron bond between our Barth boys and young men and these extraordinary triathletes.

(Cont'd on page 14)

A Decade of BSF's Extraordinary Contributors

(Cont'd from page 13)



**Barth Syndrome
Foundation**

2000 – 2010

...A decade of community

...A decade of education

...A decade of discovery

There are many more Barth ambassadors, some of whom are listed here: people like Lynda Sedefian, who led the very first Barth walk-a-thon in Voorheesville, NY in 2001; Mary, Rosemary and Ted Baffa who introduced BSF to Mrs. Annenberg and to the Lebensfeld Foundation and whose friends held a wonderful testimonial fund raising dinner in their honor; Steve and Jan Kugelmann who have held five Barth Syndrome Driving-for-a-Cure golf tournaments in Merritt Island, FL; and Randy and Leslie Buddemeyer who have held five of their own golf tournaments also in Florida to support BSF and the Juvenile Diabetes Research Foundation. The Dunns and the Monahans have been raising funds and awareness around Boston for a decade. The Bowen, Oldewage and Telles families, among others, continue their active support of BSF even after the loss of their boys, proving that BSF is truly a family through thick and thin. Joyce Lochner, Tracy Brody, and Mary Lou Pagano have all made quilts and held raffles to raise funds for and in honor of the BSF community. The Fairchild, McCurdy, and Wilkins families use the US mail to convince their friends and neighbors to support their cause; and Shelley Bowen, who cannot seem to help but raise awareness in everything she does, everywhere she goes!

We have only touched on a few of our many ambassadors here—admittedly some of the longest serving members of our little community. It is clear that a determined group of people sharing a common vision, and driven by the love of a child can be a powerful force for good. The greatest legacy we can leave this world is embodied in our children. If we are to insure that we leave them and their children with a healthy future, we must all find the courage to become ambassadors for Barth. We are where we are today because the people named above found that courage and asked their friends, neighbors, colleagues and family to support the Barth Syndrome Foundation. What will we write about in the next decade? As it was in the past, that will depend on us.

Looking to a New Decade of Discovery

A decade is a long time... unless you are trying to cure a complex genetic disorder. Just as it takes a community to raise a child... it takes a community to cure a disease. We depend on you, each a part of our community—our scientists, our physicians, our volunteers and staff, our donors and our families—to all pitch in and help, each in his or her own way. The people we honor for their service over the last decade are all extraordinary contributors. Over the next ten years, we will need all of you and others, to reach our goal of discovering more effective treatments and one day a cure. Thank you so much for your help so far; we count on you going forward!



Candids of BTHS families at BSF's 2010 conference. (Photos courtesy of Cherie Schrader.)



Awareness of Barth Syndrome is Growing Exponentially

There has been a significant increase in Barth syndrome (BTHS) related peer-reviewed journal articles published. To date, there have been **52** articles published on BTHS research conducted with the support of BSF and/or BSF affiliate funding (denoted below with an asterick). 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.

1. Acehan D, Vaz F, Houtkooper RH, James J, Moore V, Tokunaga C, Kulik W, Wansapura J, Toth MJ, Strauss A, Khuchua Z. **Cardiac and skeletal muscle defects in a mouse model of human Barth syndrome.** *J Biol Chem.* 2010 Nov 9. [Epub ahead of print] (*PubMed Abstract*)*
2. Aljishi E, Ali F. **Barth Syndrome: An X-linked Cardiomyopathy with a Novel Mutation.** *Indian J Pediatr.* 2010 Oct 28. [Epub ahead of print]. (*PubMed Abstract*)
3. Kim J, Hoppel CL. **Monolysocardiolipin: improved preparation with high yield.** *J Lipid Res.* 2010 Oct 19. [Epub ahead of print] (*PubMed Abstract*)
4. Scherer M, Schmitz G, Liebisch G. **Simultaneous Quantification of Cardiolipin, Bis(monoacylglycerol)phosphate and their Precursors by Hydrophilic Interaction LC-MS/MS Including Correction of Isotopic Overlap.** *Anal Chem.* 2010 Oct 14. [Epub ahead of print]. (*PubMed Abstract*)
5. Hershberger RE, Morales A, Siegfried JD. **Clinical and genetic issues in dilated cardiomyopathy: A review for genetics professionals.** *Genet Med.* 2010 Sep 22. [Epub ahead of print] (*PubMed Abstract*)
6. Gebert N, Ryan MT, Pfanner N, Wiedemann N, Stojanovski D. **Mitochondrial protein import machineries and lipids: A functional connection.** *Biochim Biophys Acta.* 2010 Aug 7. [Epub ahead of print] (*PubMed Abstract*)
7. Shemesh E, Annunziato RA, Arnon R, Miloh T, Kerker N. **Adherence to medical recommendations and transition to adult services in pediatric transplant recipients.** *Curr Opin Organ Transplant.* 2010 Jun;15(3):288-92. Review. (*PubMed Abstract*)
8. Wong, LHL, Chan FWK, Wong FYY, Wong ELY, Huen KF, Yeoh EK, Fok TF. **Transition Care for Adolescents and Families With Chronic Illnesses.** *Journal of Adolescent Health, In Press, Corrected Proof, Available online 11 June 2010. (Abstract)*
9. Rapley P, Davidson PM. **Enough of the problem: a review of time for health care transition solutions for young adults with a chronic illness.** *J Clin Nurs.* 2010 Feb;19(3-4):313-23. Review. (*PubMed Abstract*)
10. From the American Academy of Pediatrics: **Policy statements--Supplemental Security Income (SSI) for children and youth with disabilities. Council on Children with Disabilities.** *Pediatrics.* 2009 Dec;124(6):1702-8. (*PubMed Abstract*)
11. Christie D, Viner R. **Chronic illness and transition: time for action.** *Adolesc Med State Art Rev.* 2009 Dec;20(3):981-7, xi. Review. (*PubMed Abstract*)
12. Betz CL. **Health care transitions of youth with special health care needs: the never ending journey.** *Commun Nurs Res.* 2008 Spring;41:13-29. (*PubMed - No abstract available*)
13. Lotstein DS, McPherson M, Strickland B, Newacheck PW. **Transition planning for youth with special health care needs: results from the National Survey of Children with Special Health Care Needs.** *Pediatrics.* 2005 Jun;115(6):1562-8. (*PubMed Abstract*)
14. Steward CG, Newbury-Ecob RA, Hastings R, Smithson SF, Tsai-Goodman B, Quarrell OW, Kulik W, Wanders R, Pennock M, Williams M, Cresswell JL, Gonzalez IL, Brennan P. **Barth syndrome: an X-linked cause of fetal cardiomyopathy and stillbirth.** *Prenat Diagn.* 2010 Oct;30(10):970-6. (*PubMed Abstract*)*
15. Wilkinson JD, Landy DC, Colan SD, Towbin JA, Sleeper LA, Orav EJ, Cox GF, Canter CE, Hsu DT, Webber SA, Lipshultz SE. **The pediatric cardiomyopathy registry and heart failure: key results from the first 15 years.** *Heart Fail Clin.* 2010 Oct;6(4):401-13, vii. (*PubMed Abstract*)
16. Crimi M, Esposti MD. **Apoptosis-induced changes in mitochondrial lipids.** *Biochim Biophys Acta.* 2010 Sep 29. [Epub ahead of print] (*PubMed Abstract*)
17. Trusova VM, Gorbenko GP, Molotkovsky JG, Kinnunen PK. **Cytochrome c-lipid interactions: new insights from resonance energy transfer.** *Biophys J.* 2010 Sep 22;99(6):1754-63. (*PubMed Abstract*)
18. Kim J, Minkler PE, Salomon RG, Anderson VE, Hoppel CL. **Cardiolipin: characterization of distinct oxidized molecular species.** *J Lipid Res.* 2010 Sep 20. [Epub ahead of print] (*PubMed Abstract*)
19. Forrest CB, Bartek RJ, Rubinstein Y, Groft SC. **The case for a global rare-diseases registry.** *Lancet.* 2010 Jul 30. [Epub ahead of print] (*PubMed - No Abstract Available*)
20. Rubinstein YR, Groft SC, Bartek R, Brown K, Christensen RA, Collier E, Farber A, Farmer J, Ferguson JH, Forrest CB, Lockhart NC, McCurdy KR, Moore H, Pollen GB, Richesson R, Miller VR, Hull S, Vaught J. **Creating a global rare disease patient registry linked to a rare diseases biorepository database: Rare Disease-HUB (RD-HUB).** *Contemp Clin Trials.* 2010 Sep;31(5):394-404. Epub 2010 Jul 8. (*PubMed Abstract*)
21. Rubinstein YR, Groft SC. **Driving interest in consolidating resources for the creation of a global rare disease patient registry.** *Contemp Clin Trials.* 2010 Sep;31(5):393. Epub 2010 Jul 7. (*PubMed Abstract*)
22. Zhao W, Waisum O, Fung Y, Cheung MP. **Analysis of mitochondria by capillary electrophoresis: cardiolipin levels decrease in response to carbonyl cyanide 4-(trifluoromethoxy) phenylhydrazone.** *Eur J Lipid Sci Technol. Special Focus Issue Euro Fed Lipid Highlights Vol. 112, Iss. 9, pgs. 1058-1066, Sep 2010. (Wiley Abstract)*
23. Richesson R, Vehik K. **Patient registries: utility, validity and inference.** *Adv Exp Med Biol.* 2010;686:87-104. (*PubMed Abstract*)
24. Hasson SA, Damoiseaux R, Glavin JD, Dabir DV, Walker SS, Koehler CM. **Substrate specificity of the TIM22 mitochondrial import pathway revealed with small molecule inhibitor of protein translocation.** *Proc Natl Acad Sci U S A.* 2010 May 25;107(21):9578-83. Epub 2010 May 10. (*PubMed Abstract*)
25. Hauff K, Hatch GM. **Reduction in cholesterol synthesis in response to serum starvation in lymphoblasts of a patient with Barth syndrome.** *Biochem. Cell Biol.* 88(4): 595-602 (2010) (*PubMed Abstract*)*
26. van Gestel RA, Rijken PJ, Surinova S, O'Flaherty M, Heck AJ, Killian JA, de Kroon AI, Slijper M. **The influence of the acyl chain composition of cardiolipin on the stability of mitochondrial complexes; an unexpected effect of cardiolipin in alpha-ketoglutarate dehydrogenase and prohibitin complexes.** *J Proteomics.* 2010 Feb 10;73(4):806-14. Epub 2009 Nov 24. (*PubMed Abstract*)

Research Initiatives Relevant to Barth Syndrome

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











National Institutes of Health (NIH)	
NIAMS Small Grant Program For New Investigators (R03) Program Announcement (PAR) Number: PAR-09-031	Opening Date: January 23, 2009 Application Receipt/Submission Date(s): Multiple dates Expiration Date: October 25, 2011 http://grants.nih.gov/grants/guide/pa-files/PAR-09-031.html
<p><u>Purpose:</u> The Division of Musculoskeletal Diseases of the NIAMS supports fundamental research in bone, muscle and connective tissue biology as well as research aimed at improving the diagnosis, treatment, and prevention of diseases and injuries of the musculoskeletal system and its component tissues. Key public health problems addressed by this research include osteoporosis, osteoarthritis, orthopaedic disorders and injuries, including sports medicine and regenerative medicine and the muscular dystrophies. (This is an RO3 grant program which is designed to help young investigators.)</p>	
Pilot and Feasibility Clinical Research Grants in Diabetes, Endocrine and Metabolic Diseases (R21) Program Announcement (PA) Number: PA-09-133	Opening Date: May 16, 2009 Letters of Intent Receipt Date(s): N/A Application Receipt/Submission Date(s): Multiple dates Expiration Date: May 8, 2012 http://grants.nih.gov/grants/guide/pa-files/PA-09-133.html
<p><u>Purpose:</u> This FOA, issued by National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) and the Office of Dietary Supplements (ODS) of the National Institutes of Health, encourages exploratory/developmental clinical research related to the prevention or treatment of diabetes, obesity and endocrine and genetic metabolic diseases. The Pilot and Feasibility Clinical Research Grants Program is designed to allow initiation of exploratory, short-term clinical studies, so that new ideas may be investigated without stringent requirements for preliminary data. The short-term studies should focus on research questions that are likely to have high clinical impact. They can include testing a new prevention strategy, a new intervention, or unique combinations of therapies. A high priority is the use of such studies to help stimulate the translation of promising research developments from the laboratory into clinical practice in diabetes, endocrine diseases and genetic metabolic diseases, including cystic fibrosis.</p>	
Innovative Therapies and Tools for Screenable Disorders in Newborns (R01) Program Announcement (PA) Number: PAR-10-230	Opening Date: September 5, 2010 Letters of Intent Receipt Date: 30 days prior to application due date Application Due Date: See http://grants1.nih.gov/grants/funding/submissionschedule.htm Expiration Date: September 8, 2013 http://grants.nih.gov/grants/guide/pa-files/PAR-10-230.html
<p><u>Purpose:</u> This FOA, issued by the Eunice Kennedy Shriver National Institute of Child Health and Human Development, the National Institute of Diabetes and Digestive and Kidney Disease, the National Institute of Neurological Disorders and Stroke, and the National Institute on Deafness and Other Communication Disorders encourages Research Project Grant applications from institutions/ organizations that propose research relevant to the basic understanding and development of therapeutic interventions for currently screened conditions and “high priority” genetic conditions for which screening could be possible in the near future. In this FOA, a “high priority” condition is one for which the development of an efficacious therapy would make the condition amenable to newborn screening.</p>	
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)	

BTHS Timeline — BSF ~ A Decade of Discovery

1979		Described Barth syndrome (BTHS) as X-linked recessive cardiomyopathy with abnormal mitochondria.		
1981 & 1983		Fully described BTHS as an X-linked mitochondrial disease affecting cardiac muscle, skeletal muscle and neutrophil leucocytes. <u>Barth PG</u> , Scholte HR, Berden JA, Van dK-VMJM, Luyt-Houwen IE, Van tV-KET, Van dHJJ, Sobotka-Plojhar MA. J Neur Sc 1983; 62:327-355.		
1991		Found 3-methylglutaconic aciduria to be a clinical biochemical marker for BTHS. <u>Kelley RI</u> , Cheatham JP, Clark BJ, Nigro MA, Powell BR, Sherwood GW, Sladky JT, Swisher WP. J Pediatr 1991; 119(5):738-747.		
1995		Reported that G-CSF can be used successfully to treat neutropenia in BTHS. <u>Cox GF</u> , Pulsipher M, Rothenberg M, Korson M, Kelley RI. Am J Hum Genet 1995; 57(Suppl):A177.		
1996		Discovered tafazzin (<i>TAZ</i>) gene on the distal arm of Xq28. Bione S, D'Adamo P, Maestrini E, Gedeon AK, Bolhuis PA, <u>Toniolo D</u> . Nat Genet 1996; 12(4):385-389.		
1997		Documented heart transplantation as being successful in BTHS patient. Adwani SS, Whitehead BF, Rees PG, Morris A, Turnbull DM, Elliott MJ, de Leval MR. Pediatr Cardiol 1997; 18(2):143-145.		
1998		Shown that female carriers of BTHS are asymptomatic due to X-chromosome inactivation. <u>Orstavik KH</u> , Orstavik RE, Naumova AK, D'Adamo P, Gedeon A, Bolhuis PA, Barth PG, Toniolo D. Am J Hum Genet 1998; 63(5):1457-1463.		
1999		Discovered higher-than-expected unrelated BTHS cases in one hospital in Bristol, UK, indicating an under-diagnosis. Cantlay AM, Shokrollahi K, Allen JT, Lunt PW, Newbury-Ecob RA, <u>Steward CG</u> . J Pediatr 1999 Sep;135(3):311-5. Erratum in : J Pediatr 2000 Jun;136(1):136.		
2000		Held first BTHS Family Gathering. June 2000; Baltimore, MD. (Barth Syndrome Family Network) <u>Sue Wilkins</u> , <u>Shelley Bowen</u> , <u>Anna Dunn</u>		
	www.barthsyndrome.org	Obtained domain for Barth Syndrome Foundation, Inc.		
		Barth Syndrome Foundation, Inc. officially incorporated.		
		Established BSF Board of Directors and elected Shelley Bowen, Steve McCurdy, Kate McCurdy, Sue Wilkins and Anna Dunn.	(cont'd) 2000	
		Demonstrated that tafazzin is involved in cardiolipin remodeling in BTHS. <u>Vreken P</u> , Valianpour F, Nijtmans LG, Grivell LA, Plecko B, Wanders RJA, Barth PG. Biochem Biophys Res Comm 2000; 279(2):378-382. ▼		
		Obtained 501(c)(3) status.	2001	
		Initiated the design of BSF's logo.		
		Established BSF Scientific & Medical Advisory Board.		
		Presented clinical course and treatment of neutropenia in BTHS patients. Zeidler C, Barth PG, Bonilla MA, Bolyard AA, Boxer L, Cottle T, <u>Dale DC</u> , Donadiou J, Fier C, Freedman M, Kannourakis G, Kinsey S, Liang B, Schwitzer B, Welte K, Cham B. Blood 2001; 98(11):300a.		
		Published first issue of BSF newsletter.		
		Co-sponsored BTHS Cardiomyopathy Research Study with Drs. <u>Byrnie</u> , Spencer, Thompson & Spevak.* ▼	2002	
		Created listservs for BSF community.		
		Developed relationships with ORD and NINDS (NIH).		
		Launched BSF Research Grant Program.		
		Received NIH funding in support of BSF 2002 Conference.		
		Held 1 st International Scientific, Medical & Family Conference. October 2002; Baltimore, MD.		
		Examined how BTHS neutrophils function and excluded bone marrow failure and early clearance of cells as explanations for BTHS neutropenia. <u>Kuijpers TW</u> . Eur J Pediatr 2002 Oct;161 Suppl 1:S75-82. Epub 2002 Sep 13.		

2003	Awarded 5 research grants (\$148,942).
	 Media coverage: BTHS/BSF featured in Readers Digest. Article titled "Saving Michael Bowen."
	Translated BTHS related content on website into 13 foreign languages.
	Barth Syndrome Trust (UK & Europe) established.
	BSF of Canada officially incorporated.
	Documented phospholipid abnormalities in children with BTHS. <u>Schlame M</u> , Kelley RI, Feigenbaum A, Towbin JA, Heerdt PM, Schlieble T, Wanders RJ, DiMauro S, Blanck TJ. J Am Coll Cardiol 2003 Dec 3;42(11):1994-1999. ▼
	Constructed a TAZ1 yeast mutant model. Gu Z, Valianpour F, Chen S, Vaz FM, Hakkaart GA, Wanders RJA, <u>Greenberg ML</u> . Mol Microbiol 2004 Jan; 51(1):149-158. *
2004	Awarded 5 research grants (\$173,760).
	 BSF highlighted as "model" member group by Genetic Alliance.
	First BTHS child diagnosed <i>in utero</i> .
	Joined National Health Council.
	Held 2 nd International Scientific, Medical & Family Conference, BTHS Clinic, and SMAB Meeting. July 2004; Lake Buena Vista, FL.
2005	Established Barth Syndrome Medical Database & BioRepository @ Univ. of FL.
	 BSF representative invited to participate in NHLBI Working Group - Cardiomyopathies in Children with Rare Diseases.
	Awarded 4 research grants (\$157,000).
	Suggested that only the full-length and exon 5-deleted mRNAs of the tafazzin gene are important. <u>Gonzalez IL</u> . Am J Med Genet A. 2005 May 1;134(4):409-14.*
	Described risk of serious arrhythmias and sudden cardiac death in BTHS adolescents. Spencer CT, Byrne BJ, Gewitz MH, Wechsler SB, Kao AC, Gerstenfeld EP, Merliss AD, Carboni MP, <u>Bryant RM</u> . Pediatr Cardiol. 2005 Sep-Oct;26(5):632-7. * ▼
	BTHS featured on Discovery Health Mystery Diagnosis. Episode titled "Blood Brothers."

2006	 Hired BSF Science Director, Matthew J. Toth, PhD.
	 Strauss et al., and Degli Esposti et al., independently created zebrafish knock-in models of BTHS; Strauss demonstrated that G4.5 gene is essential for normal cardiac development in zebrafish. Khuchua Z, Yue Z, Batts L, <u>Strauss AW</u> . Circ Res. 2006 Jul 21;99(2):201-8. Epub 2006 Jun 22.*
	Awarded 5 research grants (\$163,801).
	Established membership with BBB Wise Giving.
	Documented cardiac and clinical phenotype in BTHS. <u>Spencer CT</u> , Bryant RM, Day J, Gonzalez IL, Colan SD, Thompson WR, Berthy J, Redfearn SP, Byrne BJ. Pediatrics. 2006 Aug;118(2):e337-46. Epub 2006 Jul 17. * ▼
	Received funding from NHLBI and ORDR (NIH) in support of BSF 2006 Conference.
	Held 3 rd International Scientific, Medical & Family Conference, BTHS Clinic, and SMAB Meeting. July 2006; Lake Buena Vista, FL.
	Created a Drosophila (fruit fly) model of BTHS. Xu Y, Condell M, Plesken H, Edelman-Novemsky I, Ma J, <u>Ren M</u> , Schlame M. Proc Natl Acad Sci USA. 2006 Aug 1;103(31):11584-8. Epub 2006 Jul 19. *
	Total number of living individuals diagnosed with BTHS and members of BSF surpassed 100 count.
	Created Human Tafazzin (TAZ) Gene Mutation and Variation Database. Gonzalez, IL.
	Shown that BTHS-causing mutations modeled in yeast display mislocalization of the tafazzin protein. <u>Claypool SM</u> , McCaffery JM, Koehler CM. J Cell Biol 2006 Jul 31;174(3):379-390.
	Awarded 8 research grants (\$320,000).

2007		Built on prior research of 2001 and more fully documented normal verbal but lower mathematical and visual spatial skills in BTHS patients. <u>Mazzocco MMM</u> , Henry AE, Kelley RI. J Dev Behav Pediatr 2007 Feb;28(1):22-30. * ▼
		Presented testimony at Social Security Disability Hearing on Compassionate Allowances.
2008		Held SMAB+ Meeting.
		Reported on development of BTHS screening using bloodspots and HPLC tandem mass spectrometry. <u>Kulik W</u> , van Lenthe H, Stet FS, Houtkooper RH, Kemp H, Stone JE, Steward CG, Wanders RJ, Vaz FM. Clin Chem. 2008 Feb;54(2):371-8. * ▼
		Awarded 9 research grants (\$333,000).
		Held 4 th International Scientific, Medical & Family Conference, BTHS Clinic, and SMAB Meeting. July 2006; Clearwater, FL.
		Awarded first-ever Varner Award for Pioneers in Science and Medicine to <u>Peter G. Barth, MD, PhD</u> and <u>Richard I. Kelley, MD, PhD</u> .
		
		
2009		Media coverage: BTHS/BSF featured on the TODAY Show and in PARADE Magazine.
		Met all 20 Better Business Bureau Charity Standards.
		Awarded 4 research grants (\$143,978).
		Held Genetic Diagnosis of BTHS meeting with all three US CLIA labs.
		First case of BTHS female patient. Poster presented at 11 th International Congress on Inborn Errors of Metabolism Meeting. Molecular Genetics and Metabolism, Volume 98, Issues 1-2, October 2009, Pages 89-118.
		Held SMAB+ Meeting.

2010		Acknowledged BSF 10 th anniversary ... a decade of community, education, and discovery.
		Distributed tafazzin “knockdown” mouse to five laboratories for detailed analysis. (Four US labs and one in Germany.)
		Barth Syndrome Medical Database & BioRepository moved to Children’s Hospital, Boston, MA.
		BSF representative invited to serve on planning committee of ORDR Uniting Rare Disease Research: The Intersection of Patient Registries, Biospecimen Repositories, and Clinical Data Workshop.
		BSTrust received NHS funding in support of Barth Syndrome Service Clinic in Bristol, England.
		Awarded 7 research grants (\$267,672).
		Received funding from NHLBI and ORDR (NIH) in support of BSF 2010 Conference.
		Held 5 th International Scientific, Medical & Family Conference, BTHS Clinic, and SMAB Meeting. July 2010; Orlando, FL.
		Awarded 2010 Varner Award for Pioneers in Science & Medicine to <u>Daniela Toniolo, PhD</u> and posthumously to <u>Peter Vreken, PhD</u> .
		
		Documented BTHS as being devastating <i>in utero</i> and under-diagnosed as cause of male fetus loss. <u>Steward CG</u> , Newbury-Ecob RA, Hastings R, Smithson SF, Tsai-Goodman B, Quarrell OW, Kulik W, Wanders R, Pennock M, Williams M, Cresswell JL, Gonzalez IL, Brennan P. Prenat Diagn. 2010 Oct;30(10):970-6. * ▼
		BTHS clinical trial announcement made on major website (ClinicalTrials.gov). * ▼
		*Publications that acknowledge financial support contributed by BSF and/or BSF affiliates. ▼ Publications that acknowledge biological samples (and/or information) from Barth families, the Barth Syndrome Medical Database and BioRepository (BRR), and/or BSF affiliates. To download a PDF version of BTHS bibliography, go to: http://www.barthsyndrome.org/english/View.asp?x=1356 .

Barth Syndrome Trust

A Hallmark Year for Barth Syndrome in the United Kingdom and Europe

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

I thought perhaps it was just me, just a sign of getting old(er). However, there is some small consolation in the fact that even my boys are stunned by how fast the year is flying by. It seems like yesterday that we were playing in the snow. A sudden blur of time took us to the slow summer holidays when all projects were possible (but never actually done). Now suddenly we're searching for scarves and gloves thrown at the back of wardrobes, and the inevitable countdown to Christmas begins.

Like many of you, we have a long list of things we want to accomplish in the next few months. One major project is the creation of a brand new website for BST to offer the latest information to families and physicians in as many languages as possible. This new website will allow people to access information in their home language and to form bonds with other



Dr. Bev Tsai-Goodman performs echo at the Bristol clinic.

families who either live in the same region or who speak the same language. On a related note, we are delighted that families in France will be creating a new "Barth France" — a French association aimed at finding new families, raising awareness and fundraising. We wish you all every success in your new venture and are happy to join forces to share information and work together to reach our common goal.

2010 has been a hallmark year for Barth syndrome in the UK and Europe, mostly due to the start of the NHS-commissioned Barth Syndrome Service Clinic in Bristol. This project has drawn in many people and has profited from the time and investment of so many. We continue to lay the foundation and think about new and innovative ways to serve our families in the years ahead. In October, Dr. Steward and I were asked to do a joint presentation at the Genetic Alliance UK meeting in London, in which we talked about how the NHS Service came to fruition. Many other patient support groups attended the meeting and told us that they were inspired to follow in our footsteps.

mutants – US \$40,000), sponsoring the travel and accommodation costs of Nicol Clayton (NHS Barth syndrome Dietitian) and Dr. Holger Prokisch (Scientist - Germany) to BSF's International Scientific, Medical and Family Conference in Orlando, Florida, USA in July 2010.

Some of the things we have done in 2010 include: sponsoring research into Barth syndrome by Dr. Miriam Greenberg (**Perturbation of mitophagy in cardiolipin**

closer to home, the Barth Syndrome Trust has sponsored travel costs for families attending the Bristol clinic as well as overnight accommodations for European families attending the clinic. At the September clinic this year, BST sponsored a breakfast for all the families who attended (which we hope will go some way to show our appreciation for asking the Barth boys and young men to fast for a few hours the day before!). Based on feedback from families (see page 22), we plan to organise and fund a Family Day after the March clinic next year, and we look forward to seeing you there!

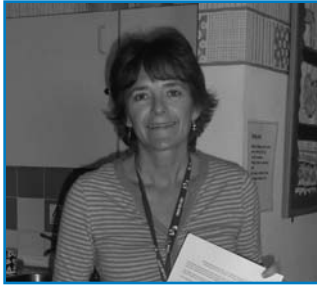
Of course, when I talk about BST sponsoring this and BST funding that, I'm really talking about you. You, our families, who make the time in an already hectic life to fundraise or who sacrifice something in order to make a regular donation to our cause. You, our volunteers; you, our friends. Thank you all... I wish you all a peaceful and happy festive season.

Playtime at the Bristol clinic held in September.



The First Barth Syndrome Service Clinic

By Debbie Riddiford, RN and Dr. Vanessa Garratt, University Hospitals Bristol NHS Foundation Trust, England



Debbie Riddiford, RN at the Bristol clinic.

The Bristol Barth Syndrome Service team were delighted to welcome 10 families to the Bristol Clinic on the 24th September 2010. Eight families came from across the UK and two came from Europe. In order to provide an in-depth service, it had been decided to have two clinic sessions a year, with some of the families attending in September and the rest in March 2011. This was the first time that many of the clinic team had met the families, and it was lovely to put names to faces and to have Michaela Damin on hand to help with introductions.

To ensure the clinic was as helpful as possible, we asked everyone to fill in a short survey before the clinic on Friday to tell us the questions that they most wanted answered during the day and the issues they wanted help with. We wanted to ensure that we talked about what mattered the most to each family. We also introduced a patient-held record for families to take to each clinic appointment so that the team could write down any advice, information and action plans as a record to take home.



Jack sits patiently during ECG at the Bristol clinic.

Families were able to hear presentations on Friday from Dr. Colin Steward (Service Lead & Haematology), Dr. Bev Tsai-Goodman (Paediatric Cardiologist), Ann Bowron (Clinical Biochemist), and Nicol Clayton (Dietitian) about what to expect in the new clinic and service. There was also feedback from BSF's 2010 international conference in Florida. Because of the expansion of the Bristol Service, all families were offered cardiac evaluations, resuscitation training, blood monitoring, and time with Dr. Steward, Dr. Tsai-Goodman, Dr. Rob Martin (Cardiologist, adolescents and transitional care), Nicol Clayton, Dr. Vanessa Garratt (Clinical Psychologist), Ruth Lumsden (Genetic Counsellor), and Tony Brooke (Dentist). A neurologist and physiotherapist were also present to offer advice and support.

On Saturday morning, Nicol Clayton was able to give feedback about managing diets and Michaela Damin, together with Debbie Riddiford (Clinical Nurse Specialist), held a focus group to evaluate the clinic (see page 22).

To all of you who attended, we would like to say a big "Thank You" for putting up with a few room changes and a lot of stairs. We really welcomed your feedback and have already changed a few elements of the clinic for March 2011. For those of you who didn't attend, we all look forward to meeting you in March.

The families would like to thank all those who organised this first Barth Syndrome Service Clinic for their hard work and commitment!

Dr. Steward (centre) and families at the Bristol clinic in September.



Families' Experiences of the Barth Bristol Clinic



Discussion group at the Bristol clinic.

Our families are at the forefront of the Barth Bristol Clinic, and so their feedback is very important and will help to shape future clinics. We started the second day of the clinic by arranging a focus group in order to discuss families' experiences of the previous day's clinic and to seek suggestions for further improvements. The feedback was very positive and the suggestions for future improvements have already been taken on board by the team and should result in an even better experience for all those attending the next clinic in March 2011. And so we mean to go on, always checking in with our families to make sure their needs are being met and always striving to change with the times and improve the service. Thank you to all the families who participated in this focus group.

“It totally exceeded all my expectations. This is the first time we've been able to talk to experts in Barth syndrome who are so knowledgeable but who are also so friendly and approachable and willing to listen to and work with us.”

Common themes and/or ideas upon which families all agreed:

How would you like the clinic to be organised?

Families thought that splitting the clinic over two days was ideal. However, their suggestion was to have an Information Day (with formal talks, informal group sessions and an emphasis on day-to-day issues) and a separate medically-orientated Clinic Day (with ECG, echo, blood tests, and consultations with specialists, etc.).

It was proposed that BST organise and fund a Family Day on the Sunday to allow families to spend time together outside the hospital environment. This would allow the children, who bonded together extremely well, to play and families to chat in an informal setting.

Was the clinic too long? Were there too many tests and was it too tiring?

Whilst families did feel it was a bit tiring, they all agreed that, since they were coming from far away, they preferred having the full array of tests, as done this year. It gave them a great sense of comfort to know that experts were looking at their child in an in-depth manner. No change necessary.

“This is the first time we've ever been able to speak to a psychologist, and I am so grateful for this opportunity.”

Did you enjoy the talks/presentations?

Yes, having the talks in the morning (rather than in the afternoon) was ideal. The talks themselves were perfectly pitched as they were informative without being overwhelming. Families want practical advice, an overview of any new developments, and a simplified summary of what is happening in research. Families also mentioned that they would like to have an additional presentation from Dr. Newbury-Ecob on the genetics of Barth syndrome.

What other parts of the clinic did you like?

Consultation with the dentist was extremely useful.

The individual colour coded appointment sheet was wonderful, as it allowed us to know exactly where we should be at what time.

“Being able to share experiences with other families is incredible—we want more!”

What could we have done differently?

Offer some sessions for the older boys, perhaps centered around a fun theme like food tasting (to try to identify which food groups are most popular as well as increase exposure to new foods).

Have feedback and/or individual appointments with the physiotherapist.

From grandparents who attended: “All the parents were united, not to complain or cry on each other's shoulders but instead to work together with the medical people to find solutions, all contributing by providing a unique perspective. They are finding comfort and hope in speaking to each other and to experts.”

“We are really happy to have been able to take part in the clinics: the meetings with you all and with the families, the clear instructive talks, the appointments with the doctors... everything was definitely worth the journey and will help us in many ways in the near future.”

More detailed feedback about the parts that worked well versus what could be improved will follow when the detailed surveys are analysed.

Jules, the First Continental European Boy to Attend the Bristol Barth Syndrome Service Clinic

By Valérie, Mother of Diagnosed Son, Belgium, Western Europe



Valérie and Jules at the 2010 Bristol clinic.

At the end of September, we crossed the Channel with our baby boy, Jules, to attend the Barth Syndrome Clinic in Bristol. This needed a bit of administrative work beforehand. We had to be in possession of Form E112 (S2) from our mutual insurance company.

In order to receive the agreement of the insurance company, Jules's cardiologist at the Clinique de l'Espérance in Liège had sent them a letter to explain why we wished to attend the clinic. Jules's team of doctors understood very well why we wanted to meet the Barth syndrome specialists and why it was important for us to go to the source of information and knowledge. This initial request was rejected by the insurance company because of a vocabulary problem. Jules's cardiologist had spoken about a "meeting" we wished to attend, while we should have specified it was a "clinic" and therefore "medical."

Our second request was thankfully successful, and it allowed Jules to attend the clinic. The two days we spent in Bristol, meeting the doctors and listening to informative and clear talks about the syndrome, really answered our expectations and were definitely worth the long journey and the administrative work.

Besides the medical part of the clinic, it was also very important for our family to be able to meet other parents in person after getting in touch over the Internet. We felt we were part of the team.

Regards from Belgium!

Families in UK and Europe — Save the Date!

Friday 25th - Saturday 26th March 2011

NHS Barth Syndrome Clinic

Bristol Royal Hospital for Children
The centre of expertise for Barth syndrome

Families from Europe must have a completed S2 form (previously E112)

followed by

Barth Syndrome Trust Family Day in Bristol

To register, please contact Barth Syndrome Trust at info@barthsyndrome.org.uk
or

Debbie Riddiford at barthsyndromeservice@uhbristol.nhs.uk

Insights from a Newly Diagnosed Family

“Strength That Comes From An Indomitable Will”

By Florence and Philippe, Parents of Newly Diagnosed Son, France



(L-R) Philippe and Florence with their three sons Romain, Raphael (BTHS), and Victor. (Photo courtesy of Amanda Clark)

Raphael is a cute 22-month old boy. Although he looks like a one-year old, he acts and understands as a boy of his age, which can be confusing sometimes.

He loves playing with his brothers, and is a very loving boy. His heart function is in the lower normal range, and looks, as many Barth syndrome (BTHS) boys, perfectly healthy. His big cheeks and his wide eyes make

people stop in the street to tell us how cute our boy is....they cannot imagine how precious he is to us....

Raphael was born on December 9, 2008. He is the third of our boys, born smaller than his brothers, Romain and Victor, but in good shape.

When Raphael was one month old, we had to go to the ER for an unexplained fever. The doctors found out that his white blood cell count was low and that something was wrong with his heart. Two months later, Raphael was diagnosed with dilated cardiomyopathy.

Cardiomyopathy, together with neutropenia, rang a bell with Raphael's cardiologist, who requested that Raphael be tested for BTHS. We had to wait five months before the results of the diagnostic test were available. This was a very long period of time, not knowing what to expect and not being able to look in any direction to get help. We felt we were stuck in the middle of nowhere.

Results from the test finally came on August 19, 2009. We will never forget this date. We couldn't consider that this diagnosis was good news. We already knew this disorder still claims the lives of many boys, but, strangely, we felt a bit relieved, since we were not alone to fight.

During all the time we were waiting for a diagnosis and also the very first days after Raphael was diagnosed, we spent a lot of time on the Internet trying to find as much information as we could. Most of the information we gained was from the Barth Syndrome Foundation (BSF) website, which was so helpful. We began to learn more about the medical aspects of the disease, and we also learned about the everyday life troubles and how to cope with low energy, gross motor delay, eating, etc.

Only a few days after we got the diagnosis, we sent an e-mail to ask how to be part of the Foundation. Shelley Bowen replied to our e-mail almost immediately and included us on BSF's Family listserv. We were also put in touch with Michaela Damin and Annick Manton of the Barth Syndrome Trust (BST). Annick is responsible for BST's family support, including families in Europe and has continued regular contact with us.

Welcoming e-mails were so warm, information was so useful, and we were amazed to see that we were not alone and that many families around the world were facing the same problems. This was such a surprise for us. It felt as if we had a new family who was able to understand what we were experiencing with Raphael.

Last January, we decided to attend BSF's 5th International Conference, which was held in July in Florida. It was, for our kids, their first “big” trip; and it was, for us, the first time we met other BTHS families who already felt to us as friends after reading their e-mails on the listserv. Even if it could be hard sometimes to meet parents that have lost their sons and to face what BTHS could mean, this conference was so full of hope. We met boys, parents, doctors, family members, all of them dedicated to one single goal: helping these boys, to which the sentence of Gandhi applies so well: **“Strength does not come from physical capacity. It comes from an indomitable will.”**

We were amazed at the commitment of those who organized the conference, and are willing to help. Back in Paris, we decided to create an Association, called “Barth France,” in order to raise money for BSF and also to raise awareness here in France. We are convinced that there are more than a couple of boys affected with BTHS in France...there are just doctors that do not know about the disease.

Philippe, his brother, Raphael's godfather and three of their friends are now training for the *Ironman France-Nice* on June 26, 2011. Inspired by Gary Rodbell's conference presentation about his fundraising and awareness success associated with Ironman races in the US, we have been in close touch ever since. We hope to be able to raise money during this race and will run under the colors of BSF.

When we realized what some mothers (the very first ones to create the Foundation) had to face, and when we measure the progress made in 10 years, we are so thankful and our hearts are full of hope. We pray that, in the near future, thanks to the commitment of families and doctors, BTHS will not claim any more lives.

Other Scientific/Medical News in Brief

UK Seeks Feedback on Rare Disease Strategy Consultation Document

In November 2008, Rare Disease UK, an alliance involving patients, clinicians, government, industry, and researchers was launched in the UK to “campaign for the adoption and implementation of national plans in each of the UK’s home nations” (England, Scotland, Wales and Northern Ireland). This past year, the five Working Groups of Rare Disease UK have been busy creating a report for the development of recommendations for the elements to include in a strategy for rare diseases. A consultation document on the initial findings of the Working Groups has now been released.

Note: The Barth Syndrome Trust is a member of Rare Disease UK.



UK Clinical Trial Opportunity: The REGENERATE-DCM Trial

An exciting series of new research studies, funded by the Heart Cells Foundation, is being conducted at Barts and The London NHS Trust to see if injecting a patient's own stem cells into their damaged heart can improve the symptoms and heart function of patients with heart failure. The REGENERATE-DCM trial started in July 2010.

60 patients will be recruited into the trial. To date, 49 have been recruited.

The trial will involve testing two different stem cell therapy techniques and therefore will have two separate arms. Both of these arms will treat 1/2 the patients with as described and 1/2 with a placebo or 'sham' procedure to remove any psychological effect from the results:

- 30 patients will receive injections of a growth factor drug (G-CSF) to stimulate the production of stem cells in the bone marrow and cause them to overspill from the marrow into the circulating blood without the need for any operation.
- 30 patients will have a five-day course of a growth factor drug (G-CSF) and then stem cells extracted from bone marrow in their hip and injected into their major coronary arteries in a minimally invasive operation.

All of the patients will be followed up with heart scans to assess the pumping function of the heart muscle to see if the treatment has caused any change.

If you suffer from heart failure and would like to know more about the study, or if you are a healthcare professional and would like to know more about the study, please visit http://www.heartcellsfoundation.com/clinical_trials.htm for further details.

Fundraising

Thank You to our Families and Friends



Suzy Green presents cheques received from their bingo fundraiser to BSTrust.

Our day-to-day activities and services for our families are supported by donations and fundraising. Recently families have been taking more and more responsibility for grassroots fundraising and raising awareness in their neighbourhoods. Thank you to all those unsung heroes who have been contributing regular amounts through standing orders for some time and to those who have recently joined their number.

We would like to thank Bernaville Nurseries of Cowley near Exeter for displaying our posters and collecting boxes during the summer and for the £107 which was collected. Thank you to Gill Amos for arranging this. Thank you to Julie Woolley and Dave Bull who continue to manage collecting boxes in their areas bringing the total this year to £1,363.31. Dave has passed the £1,000 mark for the fourth year. Thank you to Julie Woolley for her Clothing Collection fundraiser and Suzy and Jerry Green for their bingo and raffle open house evening which raised just under £800. Thank

you to a faithful friend of BST, Roger Atkins, who once again brought in much needed funds through a sponsored Rotary Charity Walk. Thank you to Sarah Whithorn for her ongoing fundraising and to her family for generously supporting her tombola (again). Sarah is hoping that the £300 raised from this will be matched by her employer.

Our families, friends, and volunteers are planning more exciting fundraising events later this year and early next year. If you want to help in a fundraising event or run your own, please get in touch with us and we'll be happy to help with ideas and advice.

Barth Syndrome Foundation of Canada

2010 A Bountiful Year for BSFCa

By Lynn Elwood, President, Barth Syndrome Foundation of Canada



Lynn, together with her son at BSF's 2010 conference. (Photo courtesy of Amanda Clark)

It is hard to believe that another year is winding down. We started this year with a brand new mission and outlook, and are working on implementing it. We are confident that as we progress with this mission, a difference will be noticed by our Canadian affected individuals and families.

It has been a busy year, especially with many of us taking part in the International Barth Syndrome Conference in Florida. Once again, we helped out with volunteer hours both before and during the conference. In addition, we provided financial assistance by sponsoring a portion of the scientific Poster Session. As well, we subsidized one of the youth activity sessions. Four Canadian physicians and scientists, namely Dr. Grant Hatch, Dr. Christopher McMaster, Dr. Richard Eband, and Dr. Mark Tarnopolsky, attended the conference and were important contributors to its success. We were very pleased to be able to sponsor three of these. We were excited to take part in such an important international endeavour.

This year we were thrilled to make new and important contacts during the conference Poster Session. We appreciated input from these individuals and their offers to help in raising awareness. Through conversations with them, we have been pointed towards possible additional contacts and organizations to partner with which will help us to grow. This will greatly enhance our ability to meet the needs of not only families who have already received a diagnosis and contacted us, but also possibly those who are still searching for solutions. We are fortunate to have volunteers in several provinces across the country whom we can depend on for assistance.



(L-R) Cathy Ritter, Lynn Elwood, Susan Hone, and Chris Hope.

We have continued to send important and relevant information to our families to include in their resource binders and are in the process of updating and revising an educator's handbook which our families will receive to give to their son's school. We know that these BSF booklets have been a great deal of help to families in the past and hope that the updated ones with Canadian specific information will be equally as useful.

Although working directly with diagnosed individuals and their families has been a primary focus this year, we are still actively running our other programs. One element that we feel is very important to many of our endeavours is our website. We have finally finished overhauling our old site and are pleased to have the new one up and running.

This site is much more user-friendly and has a fresh look; we hope it will encourage more families to contact us. We have plans to use this as a platform to boost our visibility with other charities and organizations, thereby increasing awareness in both the medical community and the general public.

We are very fortunate to have wonderful friends who not only contribute to the Foundation's fundraisers, but also take the time and effort to hold their own. Thank you to Bob and Susan McJannett for all of their work in holding a very successful and also a very fun fundraiser. With their help, our financial status remains strong. We have always tried to have a variety of events enabling us to give our supporters the opportunity to donate in a manner that is comfortable for them. One of the campaigns that we have had for several years is our annual change collection. For one month, we ask donors to collect their spare change and then donate it. While this may not be a huge money-maker, it makes it easy for people to participate, including children. Also, by keeping a jar on a kitchen counter, or even on a desk at work, it helps to bring a little bit of awareness into the community. Some donors collect their change year round. A special thank you goes out to Les and Lois for doing most of the coin rolling; in the end we took 167 rolls to the bank and each year add \$500 to \$1,000 to our finances.



Those pesky coins add up to great profits!

A Perspective From Our Volunteers

The BSFCa Annual General Meeting

By Bob McJannett, BSFCa Volunteer



Bob and Susan McJannett

My wife, Susan, and I have been supporters of BSFCa for a few years now. We were drawn in by our friends Carol, Bruce, Lois and Les. Our involvement was mostly because of our friendships with these four. It was kind of a “well, if it is important to them, it should be important to us” kind of thing. We helped out where we could, coerced our Buss Megg friends into financing the golf lunch, proved ourselves to be the dismal golfers we always thought we were, and became helpers at the golf tournament instead. Our efforts were always appreciated.

This year, because of a lack of candidates, I think, I was asked to become a Board Advisor. On April 24th, I attended the AGM, not knowing just what to expect. Well, let’s simply say I was blown away. No organization can do the right thing without interested, hard working participants. Here was a whole room

full! A dedicated, concerned group of folks, working to educate the medical community, hospital staff, etc. how to recognize Barth syndrome symptoms. Also, they provide information and support for families affected by the disorder.

But what struck me most, was the fact that everyone in the room was a volunteer, many with serious jobs that could easily distract them from their volunteering. There were only notes of hope during the meeting, descriptions of the things that had been accomplished in 2009, and an overview of projects to come.

I have friends involved with other well-financed charities, having paid staff and professional fundraisers, etc. I cannot believe they get the same feeling of satisfaction that I got knowing that the bulk of the monies BSFCa raises go directly toward BSFCa goals. I learned that other than the required expenses of auditors and insurance, the balance of your giving is either used to support the things necessary to increase donations or goes directly to research grants and the continued education of the medical profession and families.

My hat is off to everyone on the executive. You are doing a great job with little or no recognition away from that room full of volunteers. If it means anything, you have our respect and admiration. Oh, and Phyl’s butter tarts were amazing!

Vibrant Canadian Volunteers



January 9, 2010 ... cold & sunny. Canadian volunteers... warm and enthusiastic!

Very early in 2010, our good friends of the Barth Syndrome Foundation of Canada (BSFCa) gathered for a Volunteer Meeting at the home Wayne and Dianne Bridger. Our loyal and enthusiastic group once again braved the bitter cold to come together with their ideas and their yearning to help BSFCa. What a warm, loving, remarkable group of people.

Our President, Lynn Elwood, gave us a brief but engaging overview of the BSFCa’s plans for 2010. She informed everyone of the new focus for our group, our boys, our plans, and our programs. We discussed new volunteer opportunities to parallel the new direction. The group assembled were fantastic about volunteering for new jobs, brainstorming ideas, and for suggesting possible other plans.



We listened we learned! Paula, Cathy, and Marj.

We are truly enriched by our volunteers and so very thankful for their talents, their abilities, and their spirit of co-operation and willingness. Our hats go off to these very fine people. If you would like to join us / pitch in / offer your talents and experience the rush of volunteering for a fabulous cause ... please give us a shout. You can contact me directly at lois.galbraith@sympatico.ca.

From the Heart

By Maureen Pitkethly, BSFCa Volunteer



The first time I heard about Barth syndrome was from my very good friend, Audrey Hintze. She told me about losing a son at 2 ½ years and about her grandson also having the rare disease. I met her grandson and his mother, Lynn, at a Barth fundraising concert organized by Tony Murphy at the Markham Seniors' Centre.

I was very impressed with the boys, especially a small boy who played the guitar, and their enthusiasm for life even though they had this debilitating condition.

About three or four years ago, Audrey asked me if I would consider going on BSFCa's volunteer committee. Naturally, I said "sure," although I must admit that I really didn't know what I could do to help. Well, the meeting turned out to be very interesting, and when they started discussing the various areas where they needed help (such as printing and photocopying), it got my thoughts going. I realized that, although I couldn't help very much monetarily, I did have some "contacts" through having worked for the President of Xerox. Xerox was very generous and offered to cover the cost of our printing for two years, and this year they also printed a quantity of letterhead and note pads.

Both my husband, Bill, and I have helped Audrey during the poinsettia fundraiser, not only in getting orders for the plants, but also distributing them when they are delivered. Friends of mine have taken the little boxes for collecting coins. As I also volunteer at Centenary Hospital in Scarborough, I

(cont'd)

have distributed the newsletters to Cardiac and Neurological doctors there. I have mentioned Barth syndrome to many of my friends.

We are extremely impressed with the work which is done by the families of these boys and the other volunteers. Their energy seems to be endless, even though a lot of them are still working at full-time jobs. Any little way we can help, both monetarily and by the giving of our time, is a blessing to us.

“Boogie for Barth”

By Les Morris and Carol Wilks, BSFCa Volunteers

A dance was held August 21st featuring George Olliver's live music. George is a super entertainer, and the group he played to was enthusiastic and really got into the spirit of the evening. Much lively dancing and plenty of opportunity for socializing turned that part of the event into an extremely pleasant adventure. On top of everything else, a silent auction took place with lots of friendly competition among the participants who bid on an incredible selection of interesting items.

Substantial funds (\$4,268) were raised for BSFCa; and, even more importantly, an entirely new group of people was made aware of Barth syndrome. To Bob and Susan, those who purchased tickets, the band, and especially those that attended, we say "Well done!" and thank you for another successful event.

“Barth Bear Hugs” to Volunteer Celia



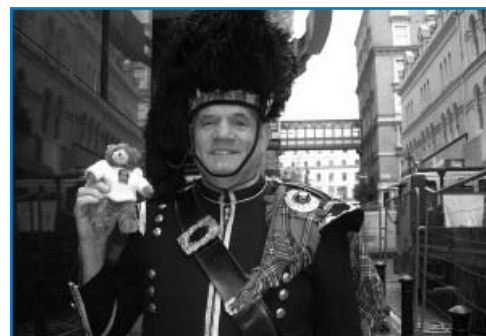
We all recognize and love the familiar BSF heart swirl! Now add that logo to a fuzzy, cuddly teddy bear and you have instant, eye-catching appeal.

Our very own Celia McGuinness has created many such beautiful Barth Bears for BSFCa.

We have distributed these Barth teddies to young Barth boys in hospitals, to our Canadian Barth gentlemen, to family members, and most recently to new Barth boys at BSF's 2010 conference in Orlando, Florida. They were a huge hit!

We thank again, Celia, and promise to bring more of the Barth Bears to the next conference.

Barth Bear visits London with Maureen.



Another Terrific Tournament BSFCa Golf Day



Josh Foursome.... (L-R) Josh, Chris, Adam, and Travis.

The spirit of golfers and supporters of BSFCa is to be commended! September in Canada for golf can be “iffy” sometimes, and this year was no exception. The days before and after our 6th Annual Golf Tournament were dampened with rains, cold and clouds; but Monday, September 13th was bright and sunny, albeit cooler by the end of our golf rounds.

This did not deter the one hundred and eight golfers or the sixteen fabulous volunteers who together made the day remarkable. This year was also blessed with the presence of five of our Barth gentlemen, Robert, Adam, Ryan, Jared and Travis. They are always crowd pleasers!

The course was extremely challenging, the day bright, the golfers enthusiastic and supportive, and the volunteers keen and smiling. The day at Tangle Creek Golf Club was a total success, and we send our thanks to the staff for all their help.

We are ever thankful to our Corporate Sponsors: Hope Aero, Ian Morris IQI, Buss Megg Society, and to the wonderful folks who attend our tournament annually because they believe in BSFCa and the commitment to push ahead with our plans and programs. Thank you, thank you, thank you!!

For information on the annual golf tournament, please contact Lois or Cathy: lois.galbraith@sympatico.ca or critter@barthsyndrome.ca



Ryan Foursome...Ryan (M), Owen, Steve, and Chris.

Heart of a Champion—Zachary Basilo



Zachary ~ April 28, 2009 – May 26, 2010

On April 28, 2009, we welcomed our second child, Zachary Michael, into the world. A few months later, we noticed some changes to Zachary's health, and, soon after, he was diagnosed with dilated cardiomyopathy. With lots of doctor appointments and medication, we now had to wait and see if his condition would improve and find out the results from genetic testing.

In December 2009, we learned that Zachary was diagnosed with Barth syndrome. As his condition continually became worse, he had to be listed for a heart transplant in January 2010. Our family moved to Toronto in order for Zachary to be on the Transplant List and so we could be close to the Hospital for Sick Children in case a donor heart arrived.

As we waited, Zachary became sick with a few cold viruses that took a toll on his body. He continued to fight hard, but his little body could only take so much. We had no other choice but for Zachary to receive a Berlin Heart (mechanic heart) to help him wait for his new donor heart. After all this, he still was the happiest boy I have ever known.

Zachary did very well on the Berlin Heart, and he was able to grow and develop mentally and physically. We enjoyed every minute with Zachary as a family at the hospital. We never knew what could happen as he had good days and bad days. We took it day by day. With the help of the Berlin Heart, our 4-year old daughter Alyssa could spend some time with him while we waited. About eight weeks after receiving the Berlin Heart, Zachary had a stroke and a few days later a major stroke. Due to these severe strokes, he had to be taken off the Transplant List at 13 months old.

Zachary, you inspired not only us but you showed so many people around the world the strength, determination and love that everyone has in them. We will always remember your smile and fight for life.

News From Barth Trust of South Africa

By Jeannette Thorpe, Chair, Barth Trust of South Africa



Jeannette and Nigel, together with their son at BSF's 2010 conference.

2010 Barth Syndrome International Conference

Once again, the Barth Syndrome International Scientific, Medical and Family Conference was an incredible success. On behalf of the Barth Trust of South Africa, I would like to extend our sincere congratulations and gratitude to all the volunteers who made this event so remarkable.

It was a great privilege to fund the attendance of Dr. Guy Letcher at this year's conference. I am tremendously grateful that he accepted the invitation to attend. He was accompanied by his wife, Caroline, who is also a nursing sister.

Dr. Letcher has recently taken over the management of the two South African Barth syndrome (BTSH) boys. He felt that it was in the best interest of these boys, and possibly others yet to be diagnosed, to learn as much as he could about BTSH.

There is no better place than our international conference! One cannot under-estimate the power of learning directly from the leading experts, being able to ask questions on the spot and, of course, opening up channels of communication. It is often extremely difficult for doctors to communicate and ask for advice from experts when they have never met them. I highly encourage any physician caring for an individual with BTSH to attend our conferences.

Furthermore, because we have so few diagnosed BTSH patients in South Africa, attending the conference gave Dr. Letcher an opportunity to meet other Barth families and individuals—from infants to young adults. Both Dr. Letcher and Caroline had an opportunity to attend and observe many of the clinics. They were extremely impressed with the calibre of this conference.

The Barth Trust of South Africa has now funded a doctor from three provinces in South Africa to attend one of BSF's international conferences.

Trustees and Plans for 2011

It was with great sadness that one of our Trustees, Carol Jardine, resigned as she has migrated to New Zealand. We wish Carol and her family all the best of luck and happiness as they start their new life.

We appointed my brother, Hunter Thyne, as the new Trustee. Hunter has a B.Com, LLB and LLM degree and specialises in Pension Fund Law. He is a highly driven man, and we are delighted to have him on board.

Our focus for 2011 will be on fundraising and awareness. In this effort, Dr. Letcher is hoping to organise an informal meeting with cardiologists to discuss the disorder.



(L-R) Dr. Richard Kelley, Dr. Guy Letcher, Caroline Letcher, and Ann Bowron at BSF's 2010 conference.



Jeannette's son with Dr. Kelley during 2010 BTSH clinic.

Sibling Spotlight

Featuring Friends from Around the Globe

Below are the profiles of two of our fantastic Barth siblings—a very important part of our Barth community.



Name: Kelsey
Age: 19
Where are you from?
New Jersey, USA
What are your hobbies?
Shopping, hanging out with friends and family.
Affected sibling? Jack, age 17

What do you like doing with your brother? I like hanging out with Jack; he's so hilarious and has a GREAT sense of humor. He always makes me laugh. I also love watching the Food Network with him.

If you met someone who had just found out that their brother had Barth syndrome, what would you say to make them feel better? Although it can be scary and stressful at times, always keep your head up and be strong for your brother. You have a huge family standing behind you, looking out for you every step of the way, and they will always be there for you.

What does the Barth Syndrome Foundation (BSF) mean to you? BSF means having a huge extended family whom you can be your complete self around and who will always be there for you, no matter what happens.



Name: Laura
Age: 19
Where are you from?
The Netherlands
What are your hobbies?
Drawing, playing games (online), hanging out with friends, and playing the guitar.
Affected sibling? Peter, age 23

What do you like doing with your brother? Playing games on the computer or just talking about things.

If you met someone who had just found out that their brother had BTHS, what would you say to make them feel better? That can be hard but that you can still have a great time with your brother. That they have to look at the things he can do and join him in those things and I'm sure that they will have a great time with him!

What does the Barth Syndrome Trust / Barth Syndrome Foundation (BST/BSF) mean to you? It helped me a lot to get to know others who are in the same situation and that we could share our experiences. I even made very good friends with other siblings and boys who have Barth syndrome.

Donations Made Easier

Donate via Check: Make check payable to **Barth Syndrome Foundation, P.O. Box 618, Larchmont, New York 10538**

Donate On-Line: You may donate to BSF or any of the international affiliates by going to our website, www.barthsyndrome.org, and clicking on the 'Support BSF' link on our home page, or through Network for Good (www.NetworkforGood.com) where donors search for BSF by name.

Donate through Causes on Facebook: Join us on our on-line social network (<http://apps.facebook.com/causes/46297/15341902>).

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

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

BSF is an accredited member of BBB



Power of Kindness



PAULA & WOODY VARNER FUND

Stars (\$5,000+)

Yanney, Michael & Dr. Gail

Angels (\$1,000 - \$4,999)

Daugherty, Robert
Wilkins, Dr. Mike & Sue

General Contributions (\$50 - \$999)

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Allman, Peter & Maureen
Allman, Tom & Jane
Basler, Dr. Rod & Debbie
Beynon, Dave & Liz
Bingham, Dr. Dave & Kathy
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Linder, Dr. Max & Pat
Massengale, Dr. Martin & Ruth
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Tegt, Dr. Tom & Barb
The Kubly Family Foundation
Varner, Judy
Varner, Tom & Beth
Wilkins, Dr. Jerry
Wilkins, Joanne
Wilkins, Marilyn
Wilkins, Dr. Lee & Kristi

SCIENCE & MEDICINE FUND

Stars (\$5,000+)

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McCurdy, Steve & Kate
McKown, Christopher (Christopher J. McKown Charitable Gift Fund)
Russell, Dr. Paul & Allene
Sernel, Marc & Tracy
The Edward C. Johnson Fund
Lummis, Frederick (The Frill Foundation)
SL 2005 Family Trust

Angels (\$1,000 - \$4,999)

Haines, Dr. Tom & Mary Cleveland
Kirkland & Ellis Foundation

General Contributions (\$50 - \$999)

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Budka, David
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Degrandis, Kathleen
Dunn, Mark & Anna
Dunn, Roland & Jean
Dunn, Stephen & Rosette
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Harrigan, Timothy & Lori

Howe, George
Katuramu, Claire
Martin, Terrence & Ellen
Laperle, Rick & Ann Marie
O'Connor, Mary
O'Brien, Dennis (Dennis B. O'Brien Land Surveying)
Ouellette, Bob & Michelle
Ouellette, Randy & Tina
Schermerhorn, Mary
Szczerpanek, Janelle
White, James & Lisa
Witzani, Gregor & Sonja

GENERAL FUND

Stars (\$5,000+)

Barad, Seth & Amy
Buddemeyer, Randy & Friends
(CB Richard Ellis 4th Annual Charity Golf Tournament)
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Janus Foundation
Lake City International Trucks, Inc.
Lummis, William & Dossy
Osnos, Peter & Susan
Pierson, Dr. Richard & Allene
Russell, Dr. Paul & Allene
Pietrini, Andrew (The Lebensfeld Foundation)

Angels (\$1,000 - \$4,999)

Alisberg, Andy & Susan
Alparo, Arlene
American Express PAC Match
Anonymous
BSF Canada
Buddemeyer, Randy & Leslie
Buly, Dr. Robert & Lynne
Cloud, Jody
Crowley, Peter & Gretchen
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Lummis, Richard & Paige
Malkin, Louisa
Malkin, Rebecca (Rebecca Swift Malkin Fund)
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Nevasier, James
North Florida Regional Thyroid Center
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Winston, Laura
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General Contributions (\$50 - \$999)

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Allen, Dr. Answorth
Allen, Tiffani
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Andrews, Sandra
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Bancroft, Rev. Francis
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Bartels, Kenneth & Jane
Barth, Andrew & Allison Demchick
Bater, Jennifer
Bell, Roberta
Bennis, Lisa
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Berger, Trisanne
Bergquist, Clay & Leslie
Berland, Jonathan & Leslie
Berns, Scott
Berry, Rick & Carla
Bez, Ann
Bhattacharyya, Chetna
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Brause, David
Braus, Paul
Brenner, Andrew & Kathleen
Brennan, Bridget
Britton, Keith
Brody, D.W. & Tracy
Brody, David & Jeanne
Brody, Jo
Brogiolo, Rocco
Brooker, Bradford & Michele
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Browning, Melvin & Helen
Bruch, Chris
Bucci, Michael & Erika
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Buddemeyer, David
Bullock, Betty
Burgess, Larry & Charlotte
Byrd, Todd & Michele
Cain, James & Rita
Calabrese, Frederick & Anne
Callahan, William & Lynn
Calvo, Jorge & Beatriz
Campana, Owen
Campbell, Anthony & Debbie
Carroll, Robert & Ann
Cassar, Yves
Castillo, Laura
Cavanaugh, Matthew & Alice
Cavanna, Dino & Barbara
Cellino, Michael & Gina
Centore, James & Sherri
Chaffin, Lynda
Chan, Shing
Chandler, Ned & Brie
Cheney, Vernon & Ellen
Chrystal, Peter & Margot
Citta, Laura
Clancy, Michael & Elyse
Clark, Randy & Bob
Clinton, Anita
Close, Gerard & Uyen
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Comer, James
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Cox, Ray & Lou
Cracchiolo, James & Marilyn
Crowley, Chuck & Diane
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Cunniffe, Bernard & Rose Marie
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Fairchild, Dewayne & Julie
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Farrell, Tim & Susan
Fast, Eric & Patricia
Faxon, Kevin & Susan
Fedich, Allison
Fernhoff, Dr. Paul
Fero, Douglas & Terry
Fields, Peter & Elyse
Fields, Wendell & Brenda
Fienberg, Meg
Fiore, Susan
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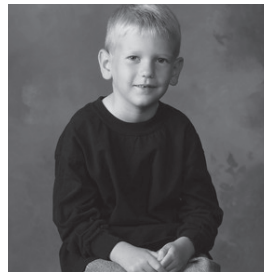
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 Coulson, Steve (Peel Plastic Products)
 Country Meat Cuts
 Cowan, Andrew
 Cowper, Margaret
 Cox, Gerald
 Cox, Patty
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 Daley, Arnold
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 Damin, Michaela
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 Davidson, Bonnie
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 Davison, Marilyn
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 Demick, Diane
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 Dickson, Bel
 DiDiodato, Giulio
 Diorkin, Gary (Peel Scrap Metal Recycling Ltd.)
 Dodds, Richard
 Dodds, Wilma
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 Doucet, Eldon
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 Elwood, Bryan
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 Elwood, Rick
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 Epand, Richard
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 Fisher, Ken (Goodrich Corporation)
 Fitzpatrick, Ann Marie
 Fitzpatrick, Bill
 Fitzpatrick, William
 Fortner, James
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 Gordon, Karen
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 Gregorio, John
 Grenc, Cathy
 Grenc, Ed
 Hackett, Doug
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 Hamilton, Glen
 Hamilton, Reg
 Hamilton, Sam
 Hamilton, Susan
 Hammand, Jean
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 Havelock District Wood Turners
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 Henders, Nola
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Hill, John
 Hill, Richard
 Hintze, Audrey
 Hocevar, Frank
 Hocevar, Helen
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 Hone, Barbara
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 Jeffery, Jeff
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 Kadoguchi, Lily
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 Kahn, Stephen
 Kahn, Vera
 Karolidis, Paul
 Kay, Jason
 Kemp, Bill
 Kennington, Paul
 King, Mark
 Kraljic, Andrew
 Kraljic, Tom
 Kritschgau, Rick (Vision 2000)
 Krueger, Rudy
 Kubien, Steve
 Kugelmann, Jan
 Kugelmann, Steve
 Kuzik, Brian
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 Lacroix, Guy
 Lajoie, Doris
 Lamb, Doug (Exceltec Auto Services)
 Lamorie, Bruce
 Lavigne, Carol
 Law, Nancy
 Lawson, Alexander S.
 Leckey, Jim
 Leighton, Jeannine
 Leighton, Keith
 Lerner, Mitchell
 Lindberg, Gunner
 Lindsay, Sue
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 Loos, Ann
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 MacDonald, Carol
 MacDonald, Fred
 MacDonald, Jody
 Macewan, Alistair
 Macewan, Chris
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 Malfara, Frank
 Mann, James
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 Marantet, Don
 Marchesin, Elvy
 Marchetti, Greg
 Marcucci, Gabe
 Marshall, Jason
 Martin Laneau, Liz
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 Masterson, Moira
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 McCurdy, Steve
 McDowell, Bill
 McDowell, Joan (J & B Customs) Brokers Ltd.
 McGill, Steve
 McGlaughlin, Larry
 McGlaughlin, Jackie
 McGuinness, Celia
 McJannett, Bob
 McJannett, Susan
 McKee, Jim
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 McLean, Cheryl
 McLean, Greg

McLennan, Alec
 McMaster, Dr. Christopher
 McMullen, Jack
 Megelink, Jasper
 Megelink, Mary
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 Mill, Dawn
 Millar, Margaret
 Millar, Ronald
 Miller, Pickard Law
 Milne, Peter
 Miloff, Michael
 Minaloff, Nick
 Mitchell, Eric
 Moe Limo
 Moffat, Heather
 Moher, Jan
 Monopoli, Domic
 Moore, Walt
 Morris, Cherniak
 Morris, Dianna
 Morris, Elsie
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 Mullen, Leatta
 Murphy, Tony (Senior Extravaganza)
 Nagel, Erna
 Nagel, Siegfried
 Nagel, Thomas
 Naraine, Christopher
 Newfeld, Beth
 Newfeld, Jeff
 Noddle, Bev
 Noddle, Norm
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 Olorenshaw, Neil
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 O'Reilly, Patrick
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 Paczkowski, Taisia
 Pape, Aaron
 Parbtani, Anwar
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 Pearce, Bev
 Pearce, William
 Perkins, Phyllis
 Perkins, Vickie
 Petzel, Sandra
 Piasecki, Jon
 Pike, Pam
 Pitkethly, Maureen
 Pixelera
 Poitras, Mike
 Poitras, Sandi
 Poole, Bob (Aerostructural Inc.)
 Post, Michael
 Power, Carolyn
 Primus, Chris
 Prodan, Magda
 Pudsey, George
 Quackenbush, Carolyn
 Quackenbush, Earl
 Ramsey, Heather
 Rantz, Rick
 Raymond, David
 Raymond, Lila
 RBC Foundation
 Reichert, Emmy
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 Ritter, Andrew
 Ritter, Carole
 Ritter, Cathy
 Ritter, Chris
 Ritter, Ryan
 Roberts, Marlowe, Jackson, Jackson & Assoc.
 Robertson, Jesse
 Robinson, Elizabeth
 Ross, Barbara
 Roy, Larry
 Royal & Sun Alliance
 Rutenberg, Karen
 Salway, Rob
 Sawh, Harold
 Sawh, Mark
 Sawh, Ruby
 Schertzl, Dawn
 Schuller, Dana
 Schuller, Dennis (Aviall Canada Ltd.)
 Scothorn, Pam
 Scott, Bob (Mx Aerospace Services)
 Scott, Debi
 Sedefian, Lynda
 Segal, Donna
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 Segal, Mona
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 Sernel, Marc
 Shaw, Blaine (Mississauga Classic) Car Club

Shepherd, Jamie (Tangle Creek Golf Club)
 Shere, Gloria
 Sheridan, John
 Sheridan, John
 Shiff, Lorne
 Sirota, Laizer
 Sisson, Lindsay
 Sisson, Natalie
 Sisson, Paula
 Skopit, Barry
 Slepkov, Martin
 Sluzar, Mark
 Smith, Chris (Tim Horton's Havelock ON)
 Smith, Christine (Tim Horton's Havelock ON)
 Smith, Glenn
 Smith, Ralph
 Smith, Ted
 Smolic, Alex
 Smolic, Joe
 Snowden, Mary
 Sorensen, Marylo
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 Stabb, Susan
 Staring, A.
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 Stewart, Cal (Havelock Foodland)
 Stitt, Barbara
 Stoll, Brian
 Stolpmann, Peter
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 Storey, Dick
 Storey, Janis
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 Sun Life Financial
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 Tarnopolski, Dr. Mark
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 Taylor, Jeff
 Taylor, Susan
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 Thomas, Brian
 Thompson, Mark
 Thorpe, Jeannette
 Timleck, Barry
 Tjart, Gwen
 Tjart, Richard
 Tocco, Carolyne (Great West Life)
 Tomlinson, Annette
 Tomlinson, Brian
 Tomlinson, Danny
 Tomlinson, Shannon
 Torrence, Erin
 Toth, Dr. Matt
 Trillium Gift of Life Network
 Trim, Norma
 Trim, William
 Tsilikas, Alana (Kelsey's 7066)
 Tsilikas, George (Kelsey's 7066)
 Turner, David
 Turner, Ron
 Tweed, Scott (Medical Post)
 Tyers, Byron
 Tyers, Robert
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 Vassallo, Jonas
 Vella, Betty
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 Weller, Sharon
 White, Leone
 Whitmore, Connie
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 Wilks, Dennis
 Wilks, Helen
 Willis, Dave
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 Young, Lenore
 Young, Ron
 Zavitz, Peter



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Do you know a boy with this genetic disorder?



Barth syndrome (BTHS; OMIM #302060)

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

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

- **Cardiomyopathy** (*usually dilated with variable myocardial hypertrophy and sometimes with left ventricular noncompaction and/or endocardial fibroelastosis*)
- **Neutropenia** (*chronic, cyclic, or intermittent*)
- **Under-developed 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 5 to 20-fold increased*)
- **Cardiolipin abnormalities** (*though currently, this can be analyzed only in a research setting*)