



Barth Syndrome
Foundation

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Barth Syndrome Journal

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

BSF International Conference Great Science, Community & Inspiration

By Adam J. Chicco, PhD, Assistant Professor, Colorado State University, Fort Collins, Colorado

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Dr. Adam Chicco

Most scientists attend international conferences and meetings where the latest advances in their fields are presented and various opportunities are provided for the exchange of ideas and networking among colleagues, trainees, and students. Such events can be very stimulating but can also be overwhelming and even a bit isolating due to the increasing breadth of topical symposia available and sheer number of attendees presenting research. As a new cardiovascular scientist in the mid-2000s, I was interested in the role that cardiolipin abnormalities play in heart disease. Finding colleagues who shared this relatively specific interest at national heart conferences consisted of scanning through sometimes hundreds of abstracts and seminar titles, often to no avail. I knew the scientists were out there somewhere, but I was clearly looking in the wrong place. I first attended a Barth Syndrome Foundation (BSF)

conference in 2010, **and wished I had been attending for years.** When asked to reflect on my experiences with these events for this newsletter, I had to find the words to describe the strong influence they have had on my research and professional life. For me, the BSF conference excels in providing three foundational aspects of any successful scientific meeting: Science, Community, and Inspiration.

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Barth Syndrome Foundation Conference An Important Chance to Connect

By Stacey, Mother of a son with Barth syndrome, Maryland



Connor (age 3), Kevin, and Stacey
(Photo courtesy of Stacey 2012)

In a sick twist of fate, we found out our son, Connor, had Barth syndrome on Valentine's Day 2012. It's also the day we joined the Barth Syndrome Foundation. Less than 24 hours later, we received a call from Shelley Bowen, and it changed our lives forever. My husband, Kevin, who spoke with Shelley Bowen, said it's a conversation he'll never forget. "I was listening to this woman, who I didn't even know, telling me she had lost two sons to this strange and scary disease and yet, at the same time, I was comforted by her voice and by her promised commitment to my family." It was remarkable.

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If You Give A Mouse A Cookie

By Lindsay Groff, Executive Director, Barth Syndrome Foundation



Lindsay and Charlotte.

(Photo courtesy of Lindsay Groff 2013)

As the mother of a three-year old, I read my fair share of children's books. One of our favorites is the contemporary classic, *If You Give a Mouse a Cookie*. It starts with a boy who gives a mouse a cookie. Then the mouse wants milk and a straw. He requests a mirror (to view his milk mustache). In the mirror, he notices he needs a haircut, so he requests scissors... and then a broom (to clean it up). This goes on and on until he gets thirsty and asks for milk which reminds him of the cookie. It is easy to relate to the idea of one thing leading to another. Recently, this concept came to mind through two dedicated parents in our community, Kristi Pena and Kevin Woodward.

Kristi Pena discovered a contest to win a professionally created video from Birds Nest Foundation. I must

confess that I harbored doubts about winning this contest. Winning depended on the number of Facebook likes, a modern day popularity contest. Given the rarity of Barth syndrome, I wondered how we could receive enough votes to beat the nationwide competition. Much to my surprise and delight, our community pulled together to receive the most votes! The founder and CEO, Avis Richards, of Birds Nest Foundation was so taken with our cause; she agreed to provide TWO videos. We created one for awareness and one for fundraising.

The annual reports had always stood out in Kevin Woodward's mind because they were still being printed with greyscale images. He felt that using full color images would look more polished and possibly draw in the reader more easily. After learning that high costs were prohibiting the Barth Syndrome Foundation (BSF) from printing certain materials in full color, the light bulb went on for Kevin. His employer produces high volume, print-on-demand investor communications and is committed to corporate citizenship. He approached his company with the request to produce the 2012 annual report at no cost to BSF, and they agreed. Kevin was right about the color images; they help bring our stories to life. In the process, it saved the Foundation considerable overhead printing costs!

In the same way that the mouse starts off with a cookie which leads to an exciting adventure, these two examples demonstrate how a simple idea can grow into something bigger. Kristi's suggestion to enter a contest led to two powerful videos. We are confident that these videos will go far in raising awareness and funds for BSF. In fact, we have already received well over 1,000 hits on YouTube! Kevin's idea about printing the annual report in full color led to significant cost savings to BSF and our most professional report to date. In addition, Kevin's company has agreed to print the 2014 conference program in full color at no cost to BSF. We have yet to see the full effects of these two initial ideas, but we have only just begun the adventure!

A Great Team Working on Our Behalf

By Marc Sernel, Chairman, Barth Syndrome Foundation



Marc Sernel

When we surveyed the Barth community last year, we asked (among many other things) how people liked the BSF newsletter. What we learned was that the newsletter was almost universally praised, with one feature clearly receiving the lowest marks — the Chairman's letter! This was certainly not an indictment of our former Chairman (more about him in a minute), but rather a comment on the typically dry material that the Chairman is supposed to write about, particularly in contrast to the much more interesting articles about the scientific research, the boys/men/families affected by Barth syndrome (BTHS), the great fundraising efforts of various people in our

(Cont'd on page 2)

A Great Team Working on Our Behalf

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community, etc. Being a person who strives to listen and learn, one of my first suggestions as Chairman was that maybe we should do away with the Chairman's letter to devote more space for other more interesting content. Unfortunately, I am also a person that strives to govern by consensus, and the consensus among the Board was that we still needed a Chairman's letter. So here it is.

The transition in becoming Chairman has been a smooth one for a lot of reasons, but none more important than the foundation that was laid by our founding and former Chairman, Steve McCurdy. I have joked that I'm not sure it would be possible to genetically engineer a better founding Chairman than Steve — Harvard MBA, smart, committed, compassionate, etc. And most impressive of all, he married someone even smarter than himself who has also contributed immeasurably to the organization! We all owe a huge debt of gratitude to Steve for all he has done in helping build BSF into the life-changing organization it is today. Despite stepping down from the Board, Steve remains as committed as ever to helping the organization with his fundraising, wisdom, experience, and passion. I look forward to working with Steve as a life-long partner as we strive to reach our ultimate goal of a treatment or cure for Barth syndrome.

I also want to thank another longstanding Board member who stepped down this year, Michaela Damin. Michaela has been an incredible resource over the years as a Board member, most particularly in helping BSF become a cohesive international organization leveraging itself throughout the world. As with Steve, Michaela isn't going anywhere — she remains the Chair of the Barth Syndrome Trust in the UK and continues to help BSF in all ways that she can. Thank you, Mic, for all you have done and continue to do for our boys and young men.

Seamless transition is the sign of a strong organization, and our recent additions to the Board bode well for our continued success. Most recently, we were fortunate to add Cathy Ritter and her great perspective as a committed Barth mom, registered nurse, and Board member of the Barth Syndrome Foundation of Canada. I am always amazed at the breadth of talent and experience we have in this organization and on our Board — doctors, nurses, MBAs, rocket scientists, etc. It's an honor to be a part, and now elected leader, of this group. And it's no longer just moms and dads of those affected on the Board, and that's a good thing. Perhaps the person who brings the most wisdom to our discussions is also our youngest Board member. John Wilkins is not only wise beyond his years, but his insight as an affected individual is invaluable as we make decisions for the organization. Our Board is diverse and strong, although we continually look for the next generation of leaders and invite those interested to reach out.

In addition to the Board, I am very excited about the team we've been able to put in place over the last several years to run the organization. Everyone knows Shelley as the heart and soul of the organization, our shining star. Matt's great work as our Science Director has allowed us to clear more hurdles faster than we ever thought possible, and he's positioned us to have even greater breakthroughs in the near future. Lindsay has brought a level of professionalism and drive to the role of Executive Director that has pushed, and will continue to push, the organization to greater and greater heights. Last but not least, Lynda is our unsung hero behind the scenes that keeps everything running smoothly. Four very different people, each with their own strengths, that combine to make a great team working on our behalf. And I would be remiss if I did not acknowledge the countless hours of assistance we get from our incredible group of volunteers and the support we get from our generous donors. Little of what we do would be possible without our volunteers' and donors' immense contributions.

I look forward to continue working with the Board, the staff, and you as we all strive to reach our ultimate goal of a treatment and/or cure for Barth syndrome. There is nothing in the world that is more important to me and my family, and we thank you for your support.

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BSF International Conference

Great Science, Community & Inspiration

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[At the BSF conferences] "I have felt privileged to have learned from several scientific "all stars" who truly represent the best in their respective fields, all directing their efforts toward the common goal of solving the mysteries of Barth syndrome. Presentation of science in this environment is particularly exciting and rewarding, especially for a new investigator. The scientific sessions are clearly about how our collective efforts can be applied toward achieving a common goal, rather than showcasing one's own scientific prowess for self-advancement. This is the way great science should be communicated and applied — for all the right reasons."

~ Adam J. Chicco, PhD

Science

The study of Barth syndrome and its molecular bases requires an integration of several rigorous scientific disciplines not routinely assembled as a single unit, including lipid biochemistry, cell/membrane biology, molecular genetics, physiology, metabolism, and clinical aspects of mitochondrial disorders. Many BSF researchers, including myself, were focused on a particular aspect of these broad research areas before being brought together by a BSF conference. In attending these events, I have felt privileged to have learned from several scientific "all stars" who truly represent the best in their respective fields, all directing their efforts toward the common goal of solving the mysteries of Barth syndrome. Presentation of science in this environment is particularly exciting and rewarding, especially for a new investigator. The scientific sessions are clearly about how our collective efforts can be applied toward achieving a common goal, rather than showcasing one's own scientific prowess for self-advancement. This is the way great science should be communicated and applied — for all the right reasons.

Community

Numerous societies and associations exist to unite scientists under a common research agenda. However, many of these are too large to provide the sense of community afforded by smaller, more specialized groups. Attending the BSF conferences, despite the growing number of attendees each year, feels more like joining a family event than simply an assembly of scientists. Indeed, it is. The attendance, fundraising efforts, and ever-present contributions of Barth patients and their families that make these events possible serve as a constant reminder of why we are there. The beautiful, family-friendly venues and small group presentation/discussion format further instill a sense of community and camaraderie that stays with us long after we return to our institutions throughout the US and abroad. Such an environment fosters collaboration, friendship, and an open exchange of ideas that enhances the quality and impact of the work we do.

Inspiration

Perhaps above all, the BSF conference has inspired me to continually hone my research toward making discoveries that could have a direct impact on the treatment or cure of Barth syndrome. Scientists are driven to their fields of inquiry by a variety of factors, including intellectual curiosity, research funding programs and availability, institutional pressures, and/or the research techniques and interests of the investigator(s) they trained with. Few scientists I have met are able to pursue research based on a personal interest in targeting a specific condition impacting those close to them. While not touched personally by Barth syndrome, **I have been deeply moved by the strength, community, and persistence of the Barth patients and their families**, which permeates the BSF conference and the Foundation itself. I came to my first BSF conference with an interest in cardiolipin and heart disease, but left humbled with a newfound commitment to contributing what I can to the BSF mission of saving lives through education, advances in treatment, and finding a cure for Barth syndrome.

My sincere thanks to the Barth Syndrome Foundation and conference attendees for your scientific contributions, generous support, and inspiration over the years. I look forward to seeing you all next June in Clearwater, Florida.



Join us at the Hilton Clearwater Beach Hotel in 2014!
(Photo courtesy of Hilton Clearwater Beach Hotel 2012)

BSF Conference ~ A Chance To Connect

(Cont'd from page 1)

"...We left that conference more determined than ever to never miss another one and to become more involved in the Barth Syndrome Foundation. Dealing with such a rare and deadly disease is difficult to say the least, but knowing there is a community of knowledgeable experts and those who can provide emotional support is invaluable." ~ Stacey, Mother of Affected Individual, Maryland



Families, physicians, and volunteers at BSF's 2012 Conference.
Don't miss the opportunity to attend our 2014 Conference in Clearwater, Florida in June 2014!!! (Photo courtesy of Amanda Clark 2012)

Shortly after, the support we received snowballed. Before we knew it, we had new friends on Facebook and a listserv that we could email to try and get our numerous questions answered. We went from feeling powerless to feeling empowered.

Everyone we met told us that we needed to attend BSF's June 2012 Scientific, Medical & Family Conference in St. Petersburg, Florida. It was a no-brainer for us. We needed to do everything in our power to help Connor. Being able to meet medical professionals and other affected boys and men from around the world was something we couldn't pass up.

As the conference neared, we got a little nervous. Everyone was talking it up so much on Facebook and email that we feared we had jumped in too quickly. My husband and I aren't really "joiners," so we just didn't know if we would fit in. Our fears were alleviated minutes after our arrival at the Don Cesar hotel (in the middle of a tropical storm and no electricity no less!) The first person we met was John Wilkins. For those who don't know John, he is one of the oldest affected men to actively participate in the Foundation and is a member of the Board of Directors. He greeted us with the biggest smile and hugs, and we relaxed immediately.

Later that night, we attended the opening ceremony. Everyone around the room introduced themselves, including us, and they showed us a slideshow with pictures of all of the families. It was such an emotional, but also uplifting evening. Seeing the older kids socializing (both affected and unaffected siblings) just

confirmed that we had made the right decision to attend. To know that Connor will get a chance every two years to connect with boys just like him is, for us, the MOST rewarding part of the conference.

In the days that followed, we met and spoke with dozens of families affected by Barth syndrome. I talked to other moms about the excruciating decision of whether or not to have more children, and my husband shared beers with other dads and talked about how they handle bullying and having a son who is never going to become an athlete. It was much needed "therapy."

We also attended several medical meetings to arm ourselves with as much knowledge as we could. I found it extremely informative hearing from the parents of older affected boys about the medical and social problems they faced. It offered me a glimpse into Connor's possible future. In between, we enjoyed plenty of alone time to absorb and process everything that we were experiencing.

We left that conference more determined than ever to never miss another one and to become more involved in the Barth Syndrome Foundation. Dealing with such a rare and deadly disease is difficult to say the least, but knowing there is a community of knowledgeable experts and those who can provide emotional support is invaluable. We hope to see you at the 2014 Conference in Clearwater, Florida!

Changing the Outcome — The Benefits of Attending the BSF International Conference

By John Lynn Jefferies, MD, MPH, FACC, FAHA, Director, Cardiomyopathy and Advanced Heart Failure Programs, The Heart Institute, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio



Milosh (age 3) flirting with nurses at Cincinnati Children's Hospital.

The Barth Syndrome Foundation (BSF) has positively impacted families world-wide through education and funding directed at improving the understanding of Barth syndrome (BTHS). The BSF is directly responsible for numerous advancements in the field as well as facilitating connections between patients and families with BTHS.

The BSF International Conference is a unique opportunity for patients, families, advocates, and scientists to join together and develop united goals in advancing the care of boys and young men with BTHS. My first experience with the conference was a few years ago. I was invited to give a lecture on cardiovascular findings in BTHS. Following my lecture, a family of four introduced themselves and expressed sincere gratitude for my discussion on left ventricular noncompaction (LVNC) cardiomyopathy. They went on to explain that their son, Milosh, had LVNC with worsening myocardial dysfunction, and that they were coming to see me for cardiac management. That meeting marked the beginning of a long and successful journey between our group and the family that also increased awareness of available treatment options in the form of a peer-reviewed publication detailing a segment of his unique journey.

Milosh had progressive symptoms of heart failure, including failure to thrive and fluid overload, that required admission to the hospital. Our team decided that based on his heart function, his only chance for long-term survival would require a heart transplant. However, during his hospital course, his clinical status became more critical necessitating the need for more advanced treatment. He was receiving the highest level of medical treatment we could offer but would not survive to transplant without additional help. A decision was made with the family to pursue an option that required surgery that offered an opportunity for stability in the form of a left ventricular assist device (LVAD), a form of mechanical circulatory support. This device, the Berlin EXCOR, would do the work of the

chamber pumping blood to the body. However, during the surgery, the team recognized that the right ventricle that pumps blood to the lungs was also severely dysfunctional. If this chamber was working poorly, the LVAD therapy alone would likely be insufficient to sustain him. So, a second device was placed to support his right ventricle, a right ventricular assist device (RVAD). This was an aggressive strategy that employed cutting-edge technologies and involved specialists from many different disciplines but it was worth all of the investment. Milosh underwent successful heart transplant and was discharged home with his parents. He continues to do well.

Milosh underscores the changing landscape of cardiac management in BTHS. Heart failure is a growing epidemic world-wide. In the United States alone, there are 650,000 new cases of heart failure diagnosed annually. This has created a sense of urgency in the medical community to develop new treatment strategies to improve the quality of life in patients battling this progressive disease. Increasingly, BTHS patients are suffering from heart failure secondary to their cardiomyopathy and concomitant myocardial dysfunction. However, in the past, males with BTHS would have had limited opportunities to receive optimal cardiovascular care given limitations in available technologies as well as limited numbers of pediatric cardiovascular heart failure specialists to diagnose cardiomyopathy and manage advanced stages of heart disease. Milosh is a shining example of the possibilities in BTHS. He benefited from the most current strategies for cardiac management that, in the past, would likely not have been offered to a boy with BTHS.

Why is the BSF conference important to me? Because it brought me into contact with this amazing family and resulted in a life-saving approach. Why is the BSF conference important to the Barth syndrome community? Because by bringing all the right people together, we are changing the outcome one patient at a time.



(L-R) Ned, Milosh, Brie, and Mirabella.
(Photos courtesy of Brie & Ned 2013)

Why BSF Conferences Are So Important To Me

By Bruce "BJ" Develle, MSW, Personal Health Coach Coordinator, Humana Cares, St. Petersburg, Florida



(L-R) BJ and Ben at BSF's 2006 conference.
(Photo courtesy of BJ Develle 2006)

As a volunteer who has been working with the Barth Syndrome Foundation (BSF) since before it was incorporated, I can truly say I've been involved in many different ways in several different settings. My favorite settings, however, have been the conferences. My first conference in 2004 cemented in my mind how important these were, and, with each conference I have attended since, I find myself wanting to do more. Just as my own role at the conferences has evolved, so have the roles of the affected guys and siblings. For my first conference, I was asked to be the small group leader of several affected guys. The warning I was given, only half-jokingly, is that some of these guys had been involved in the famous "child care break out" previously. They had decided that they wanted to be more involved than just sitting around in a room playing games while their parents attended sessions.

With this in mind, for this conference, we added informative and reflective sessions for the affected guys and their siblings. We talked about symptoms, genetics, educational and social challenges, and strategies for addressing them. During free times, we saw joking around, hanging out, and sharing between all of the affected guys and their siblings. The group pointed out how important this community was to them. They expressed the sense of normalcy that they can only feel with people who "get them" without any explanation. Over the years, the programming for the affected guys and the siblings has evolved. As conference planners, we continually work to find a balance between serious and fun. At the same time, many of the original affected guys and siblings stepped up asking to work with and mentor the younger ones. The connection between these older and younger groups is powerful and cherished by both sides. The first time I was able to witness it, I found myself mesmerized. Just typing this and remembering those images, gives me chills.

This evolution continued when, at the last conference, I watched older affected guys and siblings lead groups and attend adult sessions. In addition, the older affected guys have begun to step up into more involved roles within the organization. I know that I, as well as many parents, couldn't be prouder to watch this happen at these conferences. Depending on who you ask, you will hear a variety of answers as to what makes these

conferences so amazing. Here are some of the moments that I will never forget:

- Being a human jungle gym for kids of all ages
- Witnessing closing ceremony slideshows created by the affected guys and siblings
- Watching an affected young man standing in front of a group of researchers and talking about who he is, giving a face to the research they are performing
- Facilitating a discussion between a physician and a group of affected teens, in which they were the expert, telling the doctor what worked and what didn't for them
- Listening to affected teens celebrate their accomplishments — physically, educationally and socially
- Laughing and joking with siblings and affected guys as they lounged in a pile, carefree and seemingly more comfortable than ever before
- Facilitating a discussion between a group of fathers on their experiences and feelings about raising sons with Barth syndrome
- Being there to both grieve and celebrate friends lost to Barth syndrome
- Hearing families new to the Foundation sharing their ideas and experiences about advocacy
- Getting hugs imported from around the world (and giving them too!)
- Introducing my BSF family to the newest member of my family, and watching theirs grow as well

For me, there is no question of whether I will volunteer again, as I usually sign up for the upcoming conference just as the current one is ending. My work and family both understand how much everyone in this community means to me. I hope to see you all there in June 2014.



BJ with group at BSF's 2012 conference.
(Photo courtesy of BJ Develle 2012)

Questions Only BSF Can Answer

By Sara, Mother of a son with Barth syndrome, Georgia

My newborn has just been diagnosed with Barth syndrome. I am terrified; what does this mean?

Has anyone with Barth syndrome had a successful heart transplant? Can Barth syndrome (BTSH) present in different ways, even in the same family? My brother survived without a heart transplant; can my son do the same? Do boys with BTSH bones fracture easily? Help immediately please! Child services thinks we did it.

Do our boys need to be on a preventive antibiotic? Does it help? What are your boys' ANC? How do our boys react to daily G-CSF shots? That doesn't sound fun. Has anyone been on Carnitine? Did it help? And, most recently: My husband and I want to have another child. I am a carrier. What options do we have? What have your families decided to do?

These are all questions that I have asked since my son was born, four years ago. Most are questions that our countless specialists could not answer. So, I had to find out myself. BTSH is so rare you can't simply turn to Google like it's an ordinary ear infection. There is only one place to turn — the Barth Syndrome Foundation (BSF), its members, and the compilation of research it has sponsored. The answers BSF has provided to me are priceless.



Jeremy, Jackson (age 3) and Sara.
(Photo courtesy of Sara 2013)

When my son was born in heart failure we were in shock. As a result of Shelley Bowen's pounding the pavement to educate doctors all over the world about Barth syndrome (BTSH), a doctor who had treated my brother 20 years earlier had recently contacted my mother. He told her that my brother should be tested for BTSH. This gave us a starting place for the etiology of my son's heart failure. Only twelve hours after birth, he was flown to a pediatric cardiac ICU.

It was soon confirmed that my son, Jackson, did have BTSH. Now what? We turned to the Barth Syndrome Foundation (BSF). The doctors immediately pulled up a study that had followed four boys' experience after heart transplants. They were successful. Therefore the doctors felt comfortable listing Jackson for a heart transplant. I don't know if they would have listed him without this publication due to the lack of education on the disease. Transplantable hearts are rare. Baby hearts are even more rare, and they will not list you if they don't feel it will be successful. They will give the heart to someone who has a higher likelihood of success. Clearly, this particular

bit of research is essential. Other research has been critical too, as well as contacts. This includes information from other parents who have found what works for their sons and from medical experts like Dr. Richard Kelley and Audrey Anna Bolyard.

My brother, Walker, now age 25, went into heart failure at three weeks old and again at three years old. He required daily growth hormone injections when he did not gain any height from age two to three. In Kindergarten, he was diagnosed with a learning disability. He had a biopsy when he was eight years old that confirmed a mitochondrial disorder, but, there were no answers as to how all of these were connected. Today, he is a college graduate. His body did not allow him to play sports, so he did the next best thing — a degree in Sports Management. He willingly worked very hard and persevered. He just completed an internship with a minor league baseball team. One of the duties entrusted to him was driving the World Series trophy from the airport to an event and back! He also got to dress as the team's mascot but, shockingly, he would not send me pictures, for fear of mockery! His career goal is to be the General Manager of a minor league baseball team. With his determination and ability to work hard and learn on the job, I have no doubt that he can accomplish his goal. I am such a proud big sister! As he was growing up, my mother did not have the answers that I have as a mother raising a son with BTSH. I am extremely grateful that I have somewhere to turn for answers in navigating through this strange disease. I hope that it offers a source that can help him continue to thrive.

My most recent question to BSF at large was a question that Shelley, in cahoots with my dear mother (a wonderful lady whom I strive to emulate as a graceful woman and mother of a son with BTSH), asked me to post. My husband and I have decided to have another child! Fortunately, he has put his trust in my faith and quest for information to decide

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Sara (far right) together with her parents, son, Jackson (age 3), and brother, Walker (age 25) at Walker's graduation.
(Photo courtesy of Sara ²⁰¹²)

what the best option for us will be. So, who better to gather experience from than the other families who have had to make the same decision? There were over 30 thoughtful responses. What I got from these is that there is not a good answer, but that there is incredible faith in the families with Barth syndrome, and, there is very deep love for these boys. Again, we need more research, information, and options for our families. There was a wonderful option, called MicroSort, that the FDA did not approve because they did not want American families participating in "elective family balancing" — as if our situation is at all elective.

Our boys are wonderful blessings that often 'do things their own way.' Which is why the Foundation is so helpful. I have someone to ask "How do our boys do things?" As many of the other parents report, our boys are full of personality. Somehow when the *TAZ* gene mutated, a little extra personality was inserted. They can live full lives, but research and information and hard work are essential in helping them do so. My son, who turns four today, is extraordinary. He is bouncing with personality, love, and joy. He attends preschool, has friends, and keeps us all laughing. He takes over ten doses of medicine every day, but, other than that, I feel he lives a fairly normal life. However, we live on our toes and know that we have not been promised tomorrow, so we live today at its fullest. The parents in this Foundation are extraordinary. They will do anything for their sons, but they need the information to know what to do. We need more research!

My hope is that my son will continue to lead a full life for a long time and that my future daughter will not have to search for answers, that don't yet exist, when raising her family. The Foundation, I know, will continue to find answers to help these dreams become reality.

Someone Who Really Gets You Latrobe, Pennsylvania Family Outreach



(L-R) Jack (age 3) and Connor (age 3) are watching!
(Photo courtesy of Rachel ²⁰¹³)

By Rachel, Mother of son with Barth syndrome, Pennsylvania

You know when you're with someone who really gets you, whether that be a spouse, family member or close friend, you don't feel like you need to fill the silence? Neither of you makes the effort for needless conversation; you can just let the moment be just that — the moment.

Despite their age, this is the bond that my son Jack shares with Connor who also has Barth syndrome. I observed them numerous times — both standing at the same window and looking, no, they were *watching*. Watching the vehicles travel on Route 30 in front of the hotel, watching people load and unload from their vehicles. And then, when the moment had passed, they would take off.

Although I didn't know it when I committed to hosting a family gathering, this was precisely the reason that I did it.

(Cont'd on page 10)

Someone Who Really Gets You

Latrobe, Pennsylvania Family Outreach

(Cont'd from page 9)

Don't get me wrong....We really hit the ground running. Shelley Bowen and I met with Genetics providers at Children's Hospital of Pittsburgh Friday morning to provide information on BSF. The meeting was productive; each provider left there on-board with our mission. We had fun going to dinner at a local restaurant. After dinner, we went to the hotel to enjoy an ice cream sundae bar. Each Barth boy was given a Mr. Roger's Neighborhood trolley that night. The trolleys were generously donated by the McFeely Rogers Foundation, which is located in Latrobe; Latrobe is the birth place of Fred Rogers. I was so excited to give each boy this memorable token. On Saturday, we had a beautiful day to enjoy Idlewild Park & SoakZone, the best rated children's park in the world. We enjoyed a pizza party organized by the Maksin family. On Sunday, Dr. Kelley did a presentation for us over the internet on the metabolic aspects of Barth syndrome, explaining amino acid supplementation. We learned from Dr. Kelley, and we learned from each other. We received some very nice donations, and we got wonderful press coverage. The event was really great – need I say so myself!

The highlight of the weekend, despite my hours and hours of planning, was Shelley and the children "hiding" behind the draperies of our meeting room, singing "If You're Happy and You Know It." Absolutely priceless!

We had a wonderful weekend as a Barth family. When we said our goodbyes that Sunday, (the mission of the family gathering wasn't over. Shelley Bowen and the Maksins were going to Children's Hospital of Pittsburgh to meet with Genetics and Cardiology providers on Monday.

So now, when we pass the Wingate Hotel along Route 30 in Latrobe, Elise and Jack want to know if we are stopping to see their friends.

I hope they have nice windows (and draperies) in Clearwater!



(L-R front): Jack (age 3) and Connor (age 3); (L-R back) Sam (age 13) and Wyatt (age 5) at BSF's Latrobe Family Outreach
(Photo courtesy of Rachel 2013)

Bonding at the Family Outreach

St. Louis, Missouri

By Ashley Cade, Volunteer, Webster Groves, Missouri



(L-R): Henry (age 4), Abram (age 6), John (age 31), and Noah (age 11).
(Photo courtesy of BSF 2013)

It's always fun to experience your home town through the eyes of visitors; which is exactly what our family did in June when St. Louis hosted a Regional Barth Outreach Weekend. The weekend was unseasonably cool (thank goodness) allowing us to enjoy a few of St. Louis' outdoor gems. Starting with Friday night, we gathered at the St. Louis Zoo for Jungle Boogie. While we did not see as many animals as hoped for, we did sneak in the last train ride of the day.

Saturday morning, after breakfast at the Drury Inn, we headed to Grant's Farm. Named after Ulysses S. Grant, and previously owned by the Busch Family who owned Anheuser Busch Brewing Company, the farm is home to such animals as buffalo, elephants, camels, kangaroos, donkeys, goats, peacocks, and the iconic Budweiser Clydesdales. We had gorgeous weather to walk in and through some of the horse stables, ride the tram through the deer park region of the farm, and take in a carousel ride! Perhaps the highlight was feeding the goats their milk with baby bottles! Others may argue it was the hospitality room compliments of InBev.

(Cont'd on page 11)

Bonding at the Family Outreach

St. Louis, Missouri

(Cont'd from page 10)

Saturday evening, our group enjoyed an Italian dinner at Bartolino's complete with toasted ravioli, a regional favorite. (Toasted is just a kind word for fried)! Then we car-pooled to a St. Louis institution, Ted Drewes Frozen Custard, home of the concrete (imagine a blizzard). Everyone was taken aback by the never ending, extremely large crowd waiting for their treat. Even more shocking is how quickly they move the line! While enjoying our dessert, Dr. Todd Cade and Dr. Matt Toth gave brief updates. Todd gave an update on his studies at Washington University, and Dr. Toth gave a state of the grant program and provided updates on the progress of the upcoming clinical trial with bezafibrate (for more information, see page 13).

Sunday morning, we gathered one more time for breakfast, after which our sweet reunion was headed off to their respective homes. The weekend was a really special time for renewing old friendships, making new friendships, and enjoying a relaxed time of fun with our Barth families in the mid-west!



Families bonding at the St. Louis Outreach.
(Photo courtesy of BSF 2013)

A Barth Miracle

Tenacity and Will Power Pay Off

By Andrew, Affected Individual, Florida



Andrew (age 24).
(Photo courtesy of Andrew 2013)

My story with Barth syndrome deviates slightly from the norm. My first major complication occurred when I was an infant. After an x-ray to check for pneumonia, doctors discovered I was in heart failure. I spent a week in the hospital and was placed on several cardiac medications. By age one, my heart function had improved, and I was weaned off all medications. From then until I turned nineteen, I had minimal complications with Barth syndrome. I did have pronounced muscle weakness but was still able to participate in some sports up until third grade.

I was able to attend elementary school fulltime. My attendance was normal; I consistently achieved high grades and was able to participate in most of the same activities as my other classmates. At the age of thirteen, I entered middle school. Throughout middle school I continued to do well academically and was in honors classes.

At fourteen, I received my diagnosis of Barth syndrome. My testing was prompted by the discovery that my cousin also had the disorder. Immediately after the diagnosis, and for a few years that followed, the diagnosis did not have a substantial effect on how I lived my life (though it did put me in contact with the Barth Syndrome Foundation).

I participated in the normal high school milestones such as attending homecoming, and prom, and getting my driver's license. I continued to perform well in school. By the time I had graduated high school, I had taken 11 Advanced Placement (AP) courses, and passed 10 of 11 AP exams. This allowed me to enter college with over a year's worth of credit.

(Cont'd on page 12)

A Barth Miracle

Tenacity and Will Power Pay Off

(Cont'd from page 11)

I attended college at Florida State University (Go Noles!). My freshman and sophomore years were uneventful. I lived on campus and continued to receive high grades. During the fall semester of my junior year of college, I experienced my first major medical complication since infancy. I went into heart failure and spent the end of November and beginning of December 2009 in and out of the hospital (in retrospect, with the information I currently possess, this may have been avoidable with a change to diet and added exercise). At the end of this time period, my heart function stabilized. I had several cardiac medications added to my regime, and began to take amino acid supplements. I also changed my diet, focusing on consuming large amounts of protein. These medical issues caused me to miss the end of the fall semester, and I medically withdrew for the spring. During the spring, though, I was able to complete the remaining work that I was unable to finish in the fall semester. I then re-entered college and graduated at the end of the summer magna cum laude with two majors.

I was unable to enter law school the fall following my graduation because I had missed the application deadlines due to my medical issues the previous fall. I spent my year off optimizing my diet and starting to get into an exercise routine. Both were helpful, and I still try to maintain both when law school permits.

I applied to multiple law schools around the state of Florida. I was accepted by all of them, with scholarship offers from each. I chose to enroll at the University of Florida because it is considered the best law school in the state and one of the fifty best in the country. I entered law school in 2011 and am now currently in my third and final year. I am currently ranked in the top 20% of the class, I am on a law journal, and received a book award in tax. I also work two part-time jobs. After graduation from law school, I am planning on either returning for one more year to get a Master's in Tax Law or working in the legal field.

I hope that my story demonstrates that, despite some of the medical complications and obstacles that individuals face with this disorder, they are still able to live active and fulfilling lives. I have learned many lessons from the obstacles, and they have been helpful in dealing with the stress and work of law school by giving me perspective on what is and is not important.

Lastly, I would like to comment on education. For an individual with Barth syndrome, there are some opportunities and careers that may not be available to them because of the inherent physical limitations associated with the disorder. However, education serves as the great equalizer. With minor accommodations for any physical limitations, it creates an even playing field where an individual with Barth syndrome can perform equal to or better than his peers, and it can open up a world of almost endless possibilities, where a person with Barth syndrome can pursue a career and live a fulfilling and normal life.



The Beekman 1802 Boys with Devin (age 9).
(Photo courtesy of Devin's mother 2013)

Beekman Boys Boost BSF YouTube Hits!

Devin was overjoyed to meet the Fabulous Beekman Boys at a local book signing in his hometown recently. The Beekman boys have a popular show on the Cooking Channel and won on the TV show, *The Amazing Race*. What Devin didn't know is that his mom told them all about his journey with Barth syndrome by sharing our newly created awareness video. Thanks to the "Boys" posting on Facebook for all their fans to see, we now have just shy of 1,500 hits. That's over one thousand NEW people who now know about Barth syndrome. Check out the video on our website at www.barthsyndrome.org (under the "News & Events / What's New" section) or on BSF's YouTube channel at <https://www.youtube.com/watch?v=9W2d2l0LrOQ>.

UPDATE:

Barth Syndrome Foundation Receives Orphan-Drug Status for Bezafibrate and Pursues Next Steps



Dr. Matt Toth

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

On July 24, 2013 the Barth Syndrome Foundation (BSF) received Orphan-Drug Designation of bezafibrate for the "treatment of Barth syndrome." This achievement allows BSF and researchers to focus on studying this drug to determine if it is safe and effective for Barth syndrome individuals. The next step in this process is to file an Investigational New Drug (IND) application with the Food and Drug Administration (FDA) which explains in full detail how we are going to test this drug. This lengthy and ongoing process has only been possible through the outstanding help of: the SMARTT group at the National Heart, Lung, and Blood Institute (NHLBI), the license holder of bezafibrate (Tribute Pharmaceuticals), and the healthcare professionals who are planning and will perform the clinical study (Drs. Todd Cade and Dominic Reeds). Like the movie *The Perfect Storm* (but in a good way), these three independent groups have come together with BSF to test the first drug specifically identified for treating Barth syndrome individuals. This paragraph is just a small update to the "story to be continued." As we go forward, the BSF community will have its crucial part to play. Stay tuned!

In the meantime, a study on bezafibrate and resveratrol is being developed in the UK (for more information see page 21).



Have an Idea that Might Lead to Something Bigger?

If you want to join Kristi and Kevin to help BSF with an idea that might lead to a new adventure, let us know. Contact us at bsfinfo@barthsyndrome.org today. You never know where your initial suggestion might lead us!

(See page 2)

Opportunity to Help with Barth Syndrome Research Anesthesia Research Study

Dr. Jennifer O'Flaherty of the Dartmouth-Hitchcock Medical Center of Lebanon, New Hampshire is doing a study concerning anesthesia and Barth syndrome which needs access to some of the medical records of Barth syndrome individuals. Please consider participating in this research study and contact Dr. O'Flaherty directly if you can help.


Dr. O'Flaherty is a pediatrician and an anesthesiologist who primarily practices pediatric anesthesiology. She has a particular interest in anesthesia for children with syndromes and have published a book titled *Anesthesia for Genetic, Metabolic & Dysmorphic Syndromes of Childhood*, soon to be released in its third edition. She would like to collaborate with patients and families of patients with Barth syndrome to review and analyze the anesthetic and operative experiences of as many children/young adults with Barth syndrome as possible in order to describe which anesthetics and techniques have been successful, and to identify the nature and frequency of any complications of anesthesia. The intent is to publish this data (in aggregate — no individual people or hospitals will be identified) so that anesthesiologists all over the world will have access to the information.

For more information, please visit our website at www.barthsyndrome.org (under the "News & Events / What's New" section).

Awareness of Barth Syndrome Continues to Grow

There has been a significant increase in Barth syndrome (BTHS) related peer-reviewed journal articles published. To date, a total of **73** articles have been published on BTHS research conducted with the support of BSF and/or BSF affiliate funding (denoted below with *) and publications that acknowledge biological samples and/or information from Barth families, the Barth Syndrome Registry and Repository, and/or BSF affiliates (denoted below with ▼). Listed below are articles relevant to BTHS that have been added to BSF's library since the last issue of the Barth Syndrome Journal. To view the complete bibliography on BTHS, please visit www.barthsyndrome.org.

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6. Agarwal A, Khandheria BK, Paterick TE, Treiber SC, Bush M, Tajik AJ. [Left ventricular noncompaction in patients with bicuspid aortic valve](#). J Am Soc Echocardiogr. 2013 Sep 14. doi:pii: S0894-7317(13)00629-9. 10.1016/j.echo.2013.08.003. [Epub ahead of print]
7. Norris FC, Wong MD, Greene ND, Scambler PJ, Weaver T, Weninger WJ, Mohun TJ, Henkelman RM, Lythgoe MF. [A coming of age: Advanced imaging technologies for characterising the developing mouse](#). Trends Genet. 2013 Sep 12. doi:pii: S0168-9525(13)00143-1. 10.1016/j.tig.2013.08.004. [Epub ahead of print]
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10. Unsay J, Cosentino K, Subburaj Y, Garcia-Saez A. [Cardiolipin effects on membrane structure and dynamics](#). Langmuir. 2013 Aug 20. [Epub ahead of print]
11. Jefferies JL. [Barth syndrome](#). Am J Med Genet C Semin Med Genet. 2013 Jul 10. doi: 10.1002/ajmg.c.31372. [Epub ahead of print] (Open Access)
12. Brown DA, Sabbah HN, Shaikh SR. [Mitochondrial inner membrane lipids and proteins as targets for decreasing cardiac ischemia/reperfusion injury](#). Pharmacology & Therapeutics, Available online 15 July 2013. (Abstract)
13. Monteiro JP, Oliveira PJ, Jurado AS. [Mitochondrial membrane lipid remodeling in pathophysiology: A new target for diet and therapeutic interventions](#). Prog Lipid Res. 2013 Jul 1. doi:pii: S0163-7827(13)00042-8. 10.1016/j.plipres.2013.06.002. [Epub ahead of print]
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New Website Puts Information at Your Fingertips

www.barthsyndrome.org

Thanks to the talented designers at Pixelera and a generous grant from the Max Bell Foundation, the BSF website received a well-deserved makeover. Our revamped website offers users a modern look with intuitive navigation, updated with the latest information about Barth syndrome and our services. We hope that you will enjoy browsing our new site, finding useful information each time, and that it will be yet another tool for strengthening our community. Tell us what you think at bsfinfo@barthsyndrome.org.

Funding Opportunities Relevant to Barth Syndrome Research

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

National Institutes of Health (NIH)

Improvement of Animal Models for Stem Cell-Based Regenerative Medicine (R01)

Funding Opportunity Announcement (FOA) Number: PAR-13-114

Open Date: May 5, 2013

Application Due Date(s): Standard dates apply, by 5:00 PM local time of applicant organization

Expiration Date: May 8, 2016

<http://grants.nih.gov/grants/guide/pa-files/PAR-13-114.html>

Purpose: This FOA encourages Research Project Grant (R01) applications from institutions and organizations proposing research aimed at characterizing animal stem cells and improving existing, and creating new, animal models for human disease conditions. The intent of this initiative is to facilitate the use of stem cell-based therapies for regenerative medicine. The initiative focuses on the following areas: (1) comparative analysis of animal and human stem cells to provide information for selection of the most predictive and informative model systems; (2) development of new technologies for stem cell characterization and transplantation; and (3) improvement of animal disease models for stem cell-based therapeutic applications.

Differentiation and Integration of Stem Cells (Embryonic and Induced-Pluripotent) Into Developing or Damaged Tissues (R01)

Funding Opportunity Announcement (FOA) Number: PAR-13-094

Open Date (Earliest Submission Date): May 5, 2013

Letter of Intent Due Date(s): 30 days before application due date

Expiration Date: September 8, 2016

<http://grants.nih.gov/grants/guide/pa-files/PAR-13-094.html>

Purpose: The primary focus of the FOA is to promote in vivo studies of stem cells in animal models and in humans (if applicable) to better understand how stem cells function within developing or damaged tissues. The areas of emphasis would include systematically profiling and cataloging changes at genetic and epigenetic levels that take place in stem cells and their microenvironment. The purpose is to gain in-depth knowledge of the mechanisms involved in: progressive differentiation of Embryonic Stem Cells (ESCs) into embryonic lineages, progenitor cells and specialized cell types; adult stem cells/progenitor cells during tissue regeneration and wound healing; and Induced Pluripotent Stem Cells (iPSCs) at the site of injury during stem cell therapy.

Differentiation and Integration of Stem Cells (Embryonic and Induced-Pluripotent) Into Developing or Damaged Tissues (R21)

Funding Opportunity Announcement (FOA) Number: PAR-13-095

Open Date (Earliest Submission Date): May 16, 2013

Letter of Intent Due Date(s): 30 days before application due date

Expiration Date September 8, 2016

<http://grants.nih.gov/grants/guide/pa-files/PAR-13-095.html>

Purpose: This funding opportunity is intended to encourage innovative and high risk/impact research in the area of stem cell biology, to be explored in model organisms. The research proposed under this program can explore approaches and concepts new to this area; development of new technologies; or initial research and development of data upon which significant future research may be built. The primary focus of the FOA is to promote in vivo studies of stem cells in animal models and in humans (if applicable) to better understand how stem cells function within developing or damaged tissues. The areas of emphasis would include systematically profiling and cataloging changes at genetic and epigenetic levels that take place in stem cells and their microenvironment. The purpose is to gain in-depth knowledge of the mechanisms involved in: progressive differentiation of Embryonic Stem Cells (ESCs) into embryonic lineages, progenitor cells and specialized cell types; adult stem cells/progenitor cells during tissue regeneration and wound healing; and Induced Pluripotent Stem Cells (iPSCs) at the site of injury during stem cell therapy.

Discovery of Genetic Basis of Mendelian or Monogenic Heart, Lung, and Blood Disorders (X01)

Funding Opportunity Announcement (FOA) Number: PAR-11-307

Open Date (Earliest Submission Date): September 18, 2011

Letter of Intent Due Date: September 19, 2011; April 16, 2012; April 15, 2013; and April 14, 2014

Application Due Date(s): October 18, 2011; May 14, 2012; May 14, 2013; and May 14, 2014

Expiration Date: May 15, 2014

<http://grants.nih.gov/grants/guide/pa-files/PAR-11-307.html>

Purpose: To stimulate discoveries of the genetic basis of Mendelian or monogenic disorders that significantly affect heart, lung, and blood (HLB) systems, the NHLBI invites X01 to use the genome-wide sequencing capacity of the Mendelian Disorders Genome Centers which are funded under the HG-10-016.

(Cont'd on page 16)

National Institutes of Health (NIH)	
Health Promotion for Children With Physical Disabilities Through Physical Activity and Diet: Developing An Evidence Base (R21) Funding Opportunity Announcement (FOA) Number: PAR-11-284	Open Date (Earliest Submission Date): September 16, 2011 Letter of Intent Due Date: 30 days prior to applicable receipt date Expiration Date: September 8, 2014 http://www.grants.gov/search/search.o?mode=VIEW&oppld=110713
Purpose: This Funding Opportunity Announcement (FOA) encourages that will improve our understanding of how patterns of physical activity and dietary choice affect the health and fitness of children with physical disabilities. Proposed research should account for the functional limitations of children with disabilities and their nutritional needs, as well as the physiological, psychosocial, and environmental factors that play a role in determining the health of this population.	Exploratory/Developmental Research Grant (R21) applications
Health Promotion for Children With Physical Disabilities Through Physical Activity and Diet: Developing An Evidence Base (R01) Funding Opportunity Announcement (FOA) Number: PAR-11-288	Open Date (Earliest Submission Date): September 5, 2011 Letter of Intent Due Date: 30 days prior to applicable receipt date Expiration Date: September 8, 2014 http://grants.nih.gov/grants/guide/pa-files/PAR-11-288.html
Purpose: This Funding Opportunity Announcement (FOA) encourages understanding of how patterns of physical activity and dietary choice affect the health and fitness of children with physical disabilities. Proposed research should account for the functional limitations of children with disabilities and their nutritional needs, as well as the physiological, psychosocial, and environmental factors that play a role in determining the health of this population.	Research Project Grant (R01) applications that will improve our
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)	

You Can Make A Difference Please Join Our Team Today!

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

Who are you? If you are reading this article you are likely to find your name listed on the "Power of Kindness" pages where we celebrate our donors and those who also give us the gifts of their time and advice. Last year there were over 650 of you, more than ever before, and we hope to swell the crowd again this year. Check the listing. You are likely to see names you know — we are a small but powerful community of "friends of the families." Many of you have been acknowledged here before — some of you every year since our founding over a decade ago. Some of you may be new to Barth syndrome and to the Barth Syndrome Foundation (BSF) — welcome to the family! You are certainly a friend and a supporter and we are very grateful to you! Most of you probably know a Barth family, maybe several Barth families, and you know the story of their struggle. They probably told you about BSF and the support they have found here. Your decision to donate is a personal one meant to help them... and in your giving you join a special community that cares for one another.

You Can Make A Difference

Please Join Our Team Today!

(Cont'd from page 16)

Do you know that we rely almost entirely on your generosity to fund our programs? In the last issue you heard from Dr. Matt Toth, BSF's Science Director that we have introduced larger, multi-year Development Grants of as much as \$100,000 each. We are also having success encouraging other better funded institutions (like the National Institutes of Health) to direct their funds to Barth syndrome research, but that in turn depends on BSF "upping the ante" by increasing our own investments in research and testing. The good news is that our research is accelerating into more exciting areas. The challenge is that it costs more, so we have to find ways to raise more.

The BSF Board, Lindsay Groff (BSF's Executive Director), and our staff are working hard to make sure that every dollar you send to us is invested carefully. They have stretched your donations to do more every year. BSF proudly displays the logos of the Better Business Bureau Wise Giving Alliance, and the National Health Council and meets every one of their 20 Standards of Accountability and 43 Standards of Excellence, respectively. Their certification means we run a tight (and open) ship. The accelerating pace of scientific progress and our growing and supportive community of affected families, means that we are making a positive impact. Your continued support means that we have earned your trust and confidence.

We are coming to the end of another incredible year — a time to celebrate and to take stock of our blessings and our success. May we ask you to do two things before the end of the year? First, please make your contribution to BSF. The end of the year is crucial, as we typically receive more than 50% of our total year's donations in November and December. Make sure your name is safely on the "Power of Kindness" list next year, and, second, please spread the word! Tell one person whose name is absent from the list about BSF and why you give. Tell your story. Introduce a friend or two to our community and help us find a cure faster. **You can make a difference!**



Bly (age 9).
(Photo courtesy of BSF 2013)



Don't forget to check out BSF's fundraising video!
(Photo courtesy of BSF 2013)

Giving Tuesday — Retail has its Black Friday after Thanksgiving. Small Businesses have Small Business Saturday. Internet businesses have Cyber Monday. And now charities, including BSF, have Giving Tuesday! It seems appropriate to bracket the biggest selling days of the year with Thanksgiving and Giving Tuesday. **This year, Giving Tuesday is December 3rd**, so make sure you have a little left over after all that shopping and remember your favorite charities, and for a reminder on that subject, make sure you review our new...

Fundraising Video — which you can find on our website (under the "News & Events / What's New" section) and on BSF's YouTube channel at <https://www.youtube.com/watch?v=bts4wMwK8Bk>. Thanks to parent advocate, Kristi Pena, who learned about a contest sponsored by The Birds Nest Foundation, a New York not-for-profit which creates videos for other not-for-profits like BSF. Birds Nest offered to create a short video for the charity which had the most Facebook "likes". Kristi rallied the troops and BSF won! By the time Shelley and Lindsay finished telling our story to Avis Richards, the founder and CEO of Birds Nest, she had agreed to create two six-minute videos for BSF, one for awareness and one for fundraising. These videos tell our story in a simple, emotional, and straight-forward way and make the case for joining and giving to BSF very persuasively. Screen them at your next BSF fundraising event especially when...

Matching Gifts are involved. There are literally thousands of companies which offer to match their employees' gifts to charity. If you work for such a generous company, please make sure to let BSF know and provide us with your company's matching gift form and let your office colleagues know about BSF as well. Don't leave that money on the table! We will credit both you and your company on our "Power of Kindness" pages. Matching Gifts have already helped to increase the funds raised by...

(Cont'd on page 18)

You Can Make A Difference!

Please Join Our Team!

(Cont'd from page 17)

"When I find myself trying to complete the marathon in the dark after a long day and my body wants nothing more than to rest, it's the thought of the Barth guys who feel like this every day that gives me the strength to finish."

~ Gary Rodbell, Member of Team Will



Jaime Jofre still smiling and raising funds.
(Photo courtesy of BSF 2013)



Team Will ... what an inspiring team!
(Photo courtesy of Eliza McCurdy 2013)



Blades Battling Barth Syndrome Open.
(Photo courtesy of Milosh's father 2013)

Team Will — BSF's indefatigable runners and triathletes. For the tenth straight year, on September 29th, a dozen members of Team Will plunged into Long Island Sound in New York to begin the Jardin Westchester Triathlon, known as a sprint triathlon of "only" a .9 mile swim, a 25 mile bike ride, and a 6.2 mile run. Wearing their Team Will jerseys, in honor of Will, they were easy to spot on the bike and run (not so much in the water!) and even found themselves explaining Barth syndrome to fellow runners. Clearly they are in better shape than most of us...

Ashley Cade is also in better shape than most of us, as she has just completed running for Team Will in the Nationwide Children's Hospital Columbus 1/2 Marathon on October 20th. Ashley will be running a "mere" 13.1 miles.

Also, on November 2nd, five veteran Team Will members — Coach Gary Rodbell (age 60!), Jaime Jofre, Matt Karp, Ghent Lummis, and Stefan Tunguz competed in the Florida Ironman, going the full distance (2.4 mile swim, 112 mile bike, and 26.2 mile run) in Panama City, Florida. Each one of these athletes used his participation in the race to raise funds for Barth syndrome by asking his friends and family to give financial support to BSF. As Gary says, "When I find myself trying to complete the marathon in the dark after a long day and my body wants nothing more than to rest, it's the thought of the Barth guys who feel like this every day that gives me the strength to finish."

It was also Gary who described the initial chaotic stages of the swim portion of the Ironman as "something like trying to swim in a washing machine while someone is throwing lawn furniture at you." At least our Ironmen don't also have to compete with Ned and his...

Blades Battling Barth Tournament — Ned organized the First Annual Blades Battling Barth Syndrome Open on September 14th in Dayton, Ohio, in honor of his son, Milosh. With over 80 fencers stabbing, parrying, thrusting and slashing away with Saber, Foil and Epee for over 12 hours, the competition was reportedly fierce (but bloodless, happily). Brie, Milosh's mother, and the kids ran a bakesale and took pictures on the side. Credit goes to Ned and his NKY Fencing Academy and to Will DeVan, owner of SWORD, the two co-hosts of this premier Midwest fencing event. All proceeds in excess of costs went to BSF. The success of this event for the fencers and BSF insures that there will be a Second Annual Blades Battling Barth Tournament in 2014...better start practicing! Perhaps you can pick up a used saber at...

(Cont'd on page 19)

(Cont'd from page 18)

Lindsay's Community-Wide Yard Sale — where our own Executive Director, Lindsay Groff, spent the day coordinating a community-wide sale. She pulled together over 20 participating houses in her neighborhood to host a yard sale with a portion of the proceeds dedicated to BSF. A bright sunny day brought hundreds of bargain shoppers who all learned a bit more about Barth syndrome while hunting for treasures. Hosting a community-wide yard sale is an easy way to raise funds and awareness about Barth syndrome (and sell all that “stuff” you long ago promised your spouse to get rid of!). You never know when you are going to find a well worn Willie Mays baseball glove or a Cubs hat which you can wear to Tiffini's 3rd annual...



Hey-Hey Henry Rooftop fundraiser.
(Photo courtesy of Henry's mother²⁰¹³)

Hey-Hey Henry Rooftop Fundraiser — Chicago Cubs rooftop event in honor of her son, Henry. These outings just keep getting better and better every year (unlike the Cubs). Announcements go out for a September date when the Cubs are playing at home, the venue overlooking Wrigley Field is reserved, and the people show up for a wonderful day of baseball, fundraising, and fun, and in the center of it all is Henry with his cape and stuffed animal acting as the master of ceremonies. Henry, would you mind lending your cape to John? It might improve his game at John and Liz's annual...

Bowling for Barth — For a decade now, John and Liz and their kids have hosted their annual Bowling for Barth fundraiser in honor of their son, Jack. They held it again on October 19th in Warwick, New York. You would think after all these years that John's score would have improved, but rumor has it that it's actually the entertainment value of his performance that has increased! This event has passed the test of time and still keeps on delivering fun for its loyal bowlers, and fundraising and awareness for BSF. Thank you, John and Liz, and remember to follow through! Maybe we will see the Higgins family all dressed in their bowling shirts on the front of a...



Jack's family and friends came out once again to support Bowling for Barth.
(Photo courtesy of Jack's parents²⁰¹³)

Henry Card — as their Christmas or Holiday card this year. "Henry cards" are the invention of Tiffini and named after her son, Henry, and she has offered to make them for you to send out too! Tiffini's concept is simple, personal and a proven fundraiser for BSF. She places your favorite picture on the front and an appropriate greeting that you choose on the inside. Also on the inside is a simple message about Barth syndrome and a request to make a holiday gift to BSF. A reply envelope can easily be included. Everyone reaches out to friends and family with holiday cards, why not take the opportunity to spread awareness of Barth syndrome and ask your friends to join you in supporting a cause that means so much to all of us? Many of the Barth families have committed to sending out their own "Henry cards" this year, but get your orders in soon so you can get your cards out before the Holidays come and go! To order, contact Tiffini at tiffiniallen@gmail.com.

You provide the picture and select the greeting, BSF will pick up the cost of printing, Tiffini will work her magic, and all you have to do is sign, seal, stamp and send! Demand could be high, so get your order in ASAP!

You Can Make a Difference! — As you can see, supporting BSF is a team sport! It's fun and there are lots of coaches available. If you have an idea that you would like to turn into a fundraiser for BSF, let Lindsay Groff know at Lindsay.Groff@barthsyndrome.org and she and our Development Team can spring into action to help. Please join our team today... Help us find the funds to help us find a cure!

Barth Syndrome Trust ~ Here For Families

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

This year we welcomed several new families with young sons, while some of our boys are reaching secondary school age. Amongst the most important events for the families are the clinics run by the NHS Barth Syndrome Service in Bristol in spring and autumn, and for families with older sons, entry into secondary school.

Michaela's blog: So what happens at a Barth clinic?

The clinics are a unique way to see all the specialists, learn about current research and meet other families. They start at lunchtime on a Thursday and run all day Friday. Saturday is a Social Day for families. This October my family could only attend the Friday session, which was too rushed for Nick and so next year we hope to arrive in time for appointments on the Thursday afternoon. This is how our crazy, wonderful few days away turned out:

Thursday pm

Usual rush around getting everything organised. Arrive at Bristol Premier Inn Haymarket at around 9pm. The hotel is really good for our needs – clean and comfortable and a short walk away from the hospital.

Friday morning

7:30am: In the Beefeater for breakfast and on the lookout for fellow Barth families. My boys are so excited to spot Dillon.

8:45am: First appointment. Oops, got side-tracked — so lovely to catch up with the other families as well as all the Service staff. Debbie, the Specialist Nurse, is there to make sure everyone knows where to go. Dr. Steward is great as usual, we review all Nick's recent blood counts and his medications and have a general chat about his health.

9:15am: Next door to have Nick's signal average ECG. This takes a bit longer than a normal ECG but collects more data and can detect any subtle abnormalities.

9:45am: Into the Echo room for a detailed echo and time for Nick to catch his breath and relax while I chat to a medical student about our story.

10:15am: Nick gets hooked up to a 24 hour ECG tape that will record every heart beat and tell us if we need to worry about any possible arrhythmias.

10:30am: Consultation with the cardiologist, Dr. Tsai-Goodman, who has reviewed Nick's echo and is pleased with his heart function. Whew! That's always cause to celebrate as Nick's heart function has swung quite dramatically over the years, requiring many changes in medication and almost having a heart transplant when he was a toddler. Luckily, after eight months on the list, one day his function improved dramatically.

11:00am: Nicol Clayton, our dietician, reviews Nick's food diary and gives us advice about how to maximise his nutrition and manage his weight.

11:25am: Nick's DEXA scan is in the hospital next door so we have to run. This special scan is to check his bone density and also to give us more detailed information about his percentages of fat, bone, and muscle mass.

12:30pm: Back at the Children's Hospital they are waiting to draw Nick's blood for various tests. Nick is clearly exhausted and hungry, so Lucy (physiotherapist) and Dani (occupational therapist) kindly defer his appointments until after lunch. The phlebotomist is so good at her job: Nick hardly notices the blood draw and many of the younger boys come to tell me how easy it was.

12:45 – 1:15pm: Lunch in the Playroom where we get a much needed chance to sit down and say 'Hello' to some of the other Barth families. We haven't seen some of these boys for a year, and it's incredible to see the changes that a year can bring.

1:15 – 2:00pm: Lucy assesses Nick's strength and flexibility while Dani talks about things like his wheelchair and adaptations we might find useful around the house to increase his independence. We also discuss Nick's current physio programme and his new exercise programme at our local gym (just a few weights and some swimming to start). Then Dani puts Nick through his grip strength test which he aces. He likes a challenge! While Nick and I do the physio/OT session, Marco has taken Matthew to CPR resuscitation training. Other tracks that families could choose from include genetics and a session with Dr. Garratt, our psychologist.

2:00pm: I leave Nick at the Playroom happy with the other kids, enjoying the break.

(Cont'd on page 21)



Mothers practise CPR skills.
(Photos taken for BSTrust by Samantha Holmes 2013)

"It's a real privilege to have the two-day Barth Clinic; there is information from the specialists that we can't get from our local doctors and consultants. We can discuss with them some of our fears and worries about current and future problems and plan strategies. We are able to make decisions about his medication doses together: it's the most important day of the year for us." ~ Pete, Dad of boy affected by Barth syndrome

(Cont'd from page 20)

Afternoon Session — 2:00 - 5:00 pm Bezafibrate/Resveratrol Clinical Trial

Dr. Steward gives us an update about the application for the clinical trial of bezafibrates and resveratrol and outlines some of the major hurdles the team has faced in getting this far. Families are understandably very interested to hear about this important step of trying out a medicine that might have a real impact on their child's health. By the year end, we should have more to report.

Patients Know Best (PKB) is a useful resource which is being offered to UK Barth families. PKB are designing a secure online patient portal which is owned by the patient (or parent) so that the Bristol team can upload all your personal data from the clinic into the site and you can add whatever data is relevant (for example results from your local hospital, clinic letters, scans, blood tests, a patient journal of symptoms, etc.). We are in the process of personalising this service so that it is Barth-specific. It should be ready for testing in the next few months.

Research Study into Exercise and Metabolism

It was a great pleasure to welcome Prof. Todd Cade to the clinic. Todd is known to many who have attended the BSF conferences. He came to talk to us about his current research and quite a few of the UK families are keen to take part in this research. Todd has obligingly offered to work his research around the BSF conference dates in June 2014 to facilitate participation in both. There are many good reasons for helping with this study. Foremost is the real sense of satisfaction in being involved in research which may offer real insights into future treatments. The research grant covers costs like flights which is often the reason that families from outside the US find it so hard to attend the BSF conference.

The afternoon ended with an **open forum** and after that everyone made their (slightly shell-shocked but happy) way back to the hotel where we met up for an early dinner and some social time.



Scott, Julie, Dillon (age 8), and Prof. Todd Cade.

Saturday — Family Day

A leisurely breakfast and then we make our way to @Bristol, a brilliant interactive Science Museum where the families are free to explore and play together. Some of the families who do not usually come to this autumn clinic have joined us, and everyone is glad to meet again. A delicious lunch in a private room is organised by Annick and it's only at around 3:30pm that we start saying our goodbyes and reluctantly begin the drive home. The Bristol Barth Clinic is always full on and we can't wait to see you all at the next one!



Moi Wei and Loong (age 1).



Families and Staff at the clinic (with Prof. Todd Cade).
(Photos taken for BSTrust by Samantha Holmes 2013)

A Gathering for Dutch-speaking Families



Dutch-speaking Family Gathering in The Netherlands.
(Photo courtesy of BSTrust 2013)

Jo van Loo reports on a most enjoyable Family Gathering at the Zuiderzee Museum in Enkhuizen in The Netherlands on June 30, 2013. This indoor and outdoor museum is dedicated to the rich heritage of the area, focusing on themes of water, crafts and communities.

Families attending from our BST community were: Fioole, Swennen/Van Langendonck, Rietveld, Dubbeldam and Van Loo. We came from The Netherlands and Belgium and we had a great time together. We would like to thank the family Dubbeldam for organising this gathering. It was good to be together again.

Step Up To Secondary School!

From Michaela, Mother of a boy with Barth syndrome

Having recently gone through the challenges of starting secondary school with both my boys (Nick, age 15, with Barth syndrome (BTHS) and Matthew, age 11, who does not have Barth syndrome), I thought this would be a good time to share a little of what we've learned along the way...

Here in the UK, the move from a small, safe primary school to a huge secondary school at the tender age of eleven can be traumatic at the best of times. As a parent, once you're satisfied that the school in question will meet your child's academic needs, you tend to focus more on what will make them happy. Will they make new friends? Will they settle into the new routine where so much more is expected from them? When you have a child with complex needs like our boys with Barth syndrome, a little advance planning is crucial to ensure that the transition happens as smoothly as possible.

All our children are different and perhaps our experiences will differ from yours, but here are some of the things we've learned:

Be proactive and start planning for secondary school when your son is in Year 4-5. Identify his individual special needs (electric wheelchair, reduced timetable, special equipment, medicines, feeds, Learning Support Assistant to help him, etc.) and make sure there is a plan in place to meet them. Get a Statement of Special Needs or an IEP (Individual Education Plan) if appropriate. Check out our BST Education Guide (under Resources on www.barthsyndrome.org.uk), which has loads of useful information.

Check out the reports of your local schools before deciding where to send your son. What suits one child might not suit

another. Moving up to a secondary school where he already has some friends or a sibling may provide the security he needs for the transition. On the other hand, some families choose to send their son with Barth syndrome to a different school because of things like wheelchair accessibility, attitude of staff, concerns over bullying, etc. Some schools have specialist funding for children with physical difficulties and often can provide additional support which is invaluable.

Involve your boys in the decisions wherever possible and address their concerns. One thing we did with both our boys was reassure them that we would never leave them indefinitely in a school they hated. Yes, they had to give it a good time to settle in and we would have to work at trying to solve problems as and when they arose, but, in the end, if it really wasn't working out for them, we promised to look into moving them to somewhere else.

Open up channels of communication and find one or two key people who will be available to you to talk to when you need to convey information quickly, regularly and efficiently. An open line of communication means that little problems get solved before they snowball into big problems. A quiet word with school staff when your son is especially tired or under the weather or concerned about something in particular can mean that staff are alerted to his needs and circumstances and can step in discreetly to address them.

Ask for help! The specialist nurses and psychologist at the Bristol Service are happy to talk to you and to your child's school, if needed, and sometimes a message is better received when given by them rather than by you as a parent.

Never give up. There is usually a solution out there if you can think outside the box. Never take no for an answer.

(Cont'd on page 23)



Nick, age 15, in school uniform.
(Photo courtesy of Nick's parents 2013)

What we love about Nick's school:

They are proactive and have a 'can do' attitude. This makes all the difference in the world to us. They are not afraid of Nick's condition but instead have prepared for emergencies through regular meetings with us and with him. Staff are trained to respond in an emergency and always listen to us. They are also not afraid to be honest! If he doesn't hand in his homework without good reason, he gets detention like the other kids. He is treated like any other pupil there, but he has all the extras that he needs like a wheelchair to get around the huge school grounds and learning support assistants to keep an eye and help where needed. He has extra time for exams as well as rest breaks, an adapted timetable which gives him a study period once a week as well as an early finish one day a week and a laptop with voice recognition software. These are some of the many ways in which the school works hard to accommodate him.

From Sarah, Mother of two sons with Barth syndrome

Here are my tips for the transition, based on my experiences with Kai. I do feel that things will run slightly differently for our Ashley as his needs are different from Kai's. I have to say Kai himself has made things pretty easy for me. He has what my mum would call "an old head on young shoulders." He has had no concerns at all about going into secondary school; in fact he was looking forward to it. I think he felt some reassurance because his older Year 11 brother is at the same school and will be looking out for him!



Kai, age 12, starts secondary school.
(Photo courtesy of Kai's parents 2013)

From a parent's point of view :

Arrange a meeting. I would definitely say first and most importantly do not sit back and expect things to just happen. No matter how efficient you feel your child's primary school is, make sure you know that a meeting has been arranged at the new school, before end of term, even if this means arranging it yourself (like I did).

Make sure you have an up-to-date care plan. Take several copies to the meeting so you can be sure all relevant people will have a copy. (Template available from BST.)

Ask if your son can be paired up with a friend (if one is going to the same school). Our school actually asked who Kai would most like to have in his class with him.

Ask if the school provides lockers (somewhere to keep PE kit etc so he doesn't have

to carry it in his bag all day)

Make sure you are happy, and don't be afraid to contact the school again if you are not.

He mustn't be afraid to speak up. I have told our Kai that he needs to find his voice. There are so many different teachers teaching him every day, and they will not always realise when he may be struggling. I believe our Kai will speak up if he needs help, or if he feels unwell, or if he is unable to do something.

At the end of the day, it still boils down to me feeling happy, and I won't rest until I am.

Fundraising

There has been much activity this half-year, with heroic sponsored challenges by runners, walkers, and cyclists. Our total from all donations and Suzy Green's on-going fundraising is expected to exceed £8000 by Christmas. Thank you everyone! For details of events please see our separate Fundraising News.

Barth Syndrome Foundation of Canada Ten Years of Progress

By Lynn Elwood, President, and Chris Hope, Treasurer, Barth Syndrome Foundation of Canada



Year 2006: (L-R) Karen Gordon, Chris Hope, Cathy Ritter, Lynn Elwood, Susan Hone, Lois Galbraith, and Audrey Hintze
(Photo courtesy of BSFCa 2006)

At times, it is hard to believe that the Barth Syndrome Foundation of Canada (BSFCa) has been operating for ten years. Knowing that it would not be an easy job, several Canadian families joined together during the Barth Syndrome Foundation (BSF) 2002 international conference and decided to form a Canadian affiliate. We learned a considerable amount during those first few years as we worked through incorporating the Foundation, ensuring governance was in place, and achieving our charitable status. We worked closely with the US BSF group and set up programs focused on awareness, family services, science & medicine, and charitable support as well as fundraising.

In the early years, much of our efforts went toward raising awareness within Canada. We created material that explained Barth syndrome and shared it broadly. We attended conferences, contacted hospitals, did outreach to key types of physicians, and shared information on Barth syndrome and the organization with everyone we could reach. The result of this continuing program is a strong and growing set of physicians and researchers, families and friends that are aware of and support Barth syndrome research, affected individuals, and the organization in countless ways.

Fundraising has been very important throughout our ten years and grows in significance as we strive to expand our programs to provide more family services, and further research and support for affected individuals. We started with projects like letter appeals, selling poinsettias and collecting change. Soon, it became clear that if we could raise more funds we could expand our efforts. We held our first golf tournament in 2005 and have raised over \$150,000 to date with this annual event. Throughout our history, grassroots fundraising has played a key role, bringing both funds and awareness with activities like sales, draws, parties and events put together by families, friends, and volunteers.

Our Science & Medicine program has been a goal from the beginning and really grew when we started to fund research grants in 2007. We participate in the BSF grant process which utilizes the international Scientific and Medical Advisory Board, Science Director, and several outside researchers who review and prioritize grants from around the world that further research into all aspects of Barth syndrome. We have contributed over US \$220,000 in funding to eight research grants from Canadian and international researchers. This is a very exciting aspect of the work that we do, as research has greatly increased the worldwide understanding of the disorder and allows us to get closer to recommended treatment options.

Throughout our ten years, there has been a program that directly focuses on families and individuals affected by Barth syndrome. Through this program, we work directly to educate and support families and to help individuals affected by the condition. This support has involved everything from being at the end of the phone or mail to help with concerns; providing educational material; advocating within the healthcare system; and bringing families together so that they can share experiences, fun, and laughter. As we closed our fifth year, we spent time reviewing the organization and our path forward. At that juncture, we agreed to refocus the organization with a mission that more tightly focuses on the Canadian individuals and the things they need to have for better short- and long-term outcomes. We did a needs assessment with the affected individuals and have changed our programs and mission to more closely respond to their needs.

Our revised mission is:

Enhancing the lives and outcomes of Canadian individuals and families affected by Barth syndrome.

We are grateful for the many hours put in by our growing set of volunteers. We remain a completely volunteer organization and none of what we do could be accomplished without the countless hours from so many devoted people. We now have a program that focuses on finding, engaging, and sharing information with volunteers. Working with this amazing group of people has become one of the most rewarding parts of the organization.

It is with a great deal of satisfaction that we are able to look at the boys who have become young men in the past ten years. When we started, the ages of the individuals we work for ranged mainly from six years old to the early teens. We now have several men in their late teens and upward who play a vital role in helping to raise awareness and in informing us of their needs.

(Cont'd on page 25)



As we close our first ten years and look towards the next decade, we realize that we have learned, grown, and accomplished a great deal. We welcome input and ideas to help us grow to the next level and sustain the organization in the spirit it has enjoyed during our first decade. Thank you to all of you for your part in this amazing group. We are making a difference in the lives of Canadians and others around the world who are in some way affected by Barth syndrome.

Year 2001: First Canadian Outreach

(L-R) Chris Hone, Robert, Justin, Adam, Shane, Andrew, Michael Hope, Chris Hope, Travis, Jennifer, Susan Hone, Moira Masterson, Jared, Josh, Karen Gordon, Lynn Elwood, Richard Elwood, and Shannon.

(Photo courtesy of BSFCa 2001)

Grant Hatch...Friend and Advocate of BSFCa

By Grant M. Hatch, Ph.D., Professor, Dept. of Pharmacology & Therapeutics, University of Manitoba, J. Buhler Research Centre, Faculty of Medicine, Winnipeg, Canada

"My experience at these meetings has been an inspiration to what I now do and how I approach my work. I have received several grants over the years from the Barth Syndrome Foundation and the Barth Syndrome Foundation of Canada. Based upon the funding of these grants, I have published several papers on Barth syndrome that I hope have led to a greater understanding of the disease." ~ Grant Hatch, PhD



Prof. Grant Hatch
(Photo courtesy of
BSFCa 2013)

It has been just over a decade since I received the phone call from Dr. Richard Kelley from John Hopkins University in Baltimore inviting me to come to a meeting in Baltimore focussed on cardiolipin and Barth syndrome. I was on sabbatical at the University of Minnesota when I read with interest the paper by Dr. Peter Vreken from Dr. Peter Barth's group in Holland which showed that defective cardiolipin remodelling could be involved in the pathology of Barth syndrome. Since I had just begun work on cardiolipin remodelling a couple of years back, I

was excited and very enthusiastic to visit Baltimore and meet the scientists involved in this rare but intriguing disease.

I quickly realized that this was a serious condition which impacts not only the young boys that are diagnosed with the disease but also their families. The International Scientific, Medical & Family Conferences were then developed through

the Barth Syndrome Foundation (BSF) to not only bring together doctors and scientists to discuss and collaborate in the understanding and treatment of the disease but also to bring together families to share information and provide their stories.

The meetings are truly unique from any other scientific conferences I attend, as basic scientists, such as me, can meet front line clinicians involved directly in patient care and the individuals with the disease and their families. My experience at these meetings has been an inspiration to what I now do and how I approach my work. I have received several grants over the years from the Barth Syndrome Foundation and the Barth Syndrome Foundation of Canada. Based upon the funding of these grants, I have published several papers on Barth syndrome that I hope have led to a greater understanding of the disease. I was subsequently successful in obtaining a five-year grant from the Canadian Institutes of Health Research to continue with my studies. The Barth Syndrome Foundation of Canada has been instrumental in all of my efforts.

Thank you, Dr. Hatch!!

Editor's Note: Dr. Grant Hatch is a Canadian Researcher, University of Manitoba, who has attended several of our BSF conferences and he serves on the Scientific and Medical Advisory Board of the BSF. He is an inspiration to us here in Canada. He is a great supporter of the BSFCa and is helping to promote awareness of this unique disease.

Faces to Test Tubes

By Cathy Ritter, Vice-President, and Chris Hope, Treasurer, Barth Syndrome Foundation of Canada



Cathy and Chris.
(Photo courtesy of BSCa 2013)

A big part of Barth Syndrome Foundation of Canada's mission is to raise awareness about Barth syndrome (BTHS) in the medical community, and so to that end, two Board members, Chris Hone and Cathy Ritter, travelled west to Winnipeg where Cathy made her presentation to the Manitoba Institute of Child Health.

She spoke to two broad audiences, the first including researchers, front-line physicians, paediatric specialists, lab technicians, and post docs. It was an all-inclusive presentation, starting off with her family history and then going into the many components of the disorder. One professor said that he will now include Barth syndrome in the curriculum for his students, while another researcher commented that he was reminded that his research is being done for a reason, and that she has put a face on his test tubes. All were very much in agreement that they were truly motivated to continue asking questions, and to look for answers.

At the second presentation, Cathy's audience consisted primarily of geneticists and residents. The information was moving, informational, and upbeat, keeping the audience truly attentive and involved. Although many were aware of the disorder, Cathy was able to answer questions and inform them about better diagnostic methods, and she also stressed the importance of a diagnosis. Once again, the participants were inspired and motivated. We believe that they will now be considering BTHS on a more regular basis and will test appropriately. They have assured us they will not miss the diagnosis — they almost seemed to be looking forward to their first one.

We had a very pleasant day at the Institute and cannot thank Dr. Grant Hatch enough for taking the time not only to set everything up, but for spending the day with us. We were given a wonderful tour of the facility and received an update on his current research projects. Engaging discussions about the many facets of BTHS occurred, and it was wonderful to hear his enthusiasm and multitude of ideas for future directions and possibilities of research. Projects like this are truly motivating and inspiring for all concerned.

From the Heart!

By Bob and Susan McJannett, Volunteers, BSF of Canada



Susan and Bob McJannett
(Photo courtesy of BSCa 2013)

Les Morris asked me to explain how Susan and I became involved with the BSFCa. First, you need to know that Susan and I are two of the luckiest people in the world! We have never faced serious illness, have two sons that make our chests swell with pride, a successful small business, and one of the greatest groups of friends a couple could ever ask for.

Two of our best friends are Bruce and Carol Wilks, in fact a friendship that extends over 50 years. Through Bruce and Carol, we met and became friends with Les Morris and Lois Galbraith, and, as they say, the rest is history.

Somehow we were press ganged into playing in the Barth golf tournament. Those who know us know we are not golfers; in fact, we did so poorly we embarrassed our playing partner by winning the "most honest golfer" trophy. Once we had proven our ineptitude at that silly game, Lois promoted us to golf tournament helpers.

It was during that time while working with other volunteers we learned about Barth syndrome and its effects. We met Barth boys

who soon became Barth men. We also met their families and became more aware of the trials and tribulations each faced dealing with this relatively unknown disease. At the same time, we became aware of how little funding was available for research and the huge efforts our friends and the other Barth families were putting in to raise desperately needed cash.

It seemed like something where we could make a difference. We liked the idea that all the funds raised were going to benefit the Foundation, no one was taking any salary, and expenses were being kept to a bare minimum. We started by coercing our friends in the BussMegg society to sponsor the lunch at the golf tournament; have been able to run a couple of silent auctions at one of the car shows; and operated two Boogie for Barth dances. These were all labours of love and semi successful. This year, we are looking for something different. If anyone has any ideas we are interested in hearing them. It is imperative that we keep the funds coming in.

One of my concerns for the future is the age of the hard core workers; no one is getting any younger. Who will be the next generation of fund raisers? It is time to recruit the next group to step up to the plate.

Now Les asked for about 350 words so that is what I have written. But I could have put the whole adventure into a single sentence. "It was the right thing to do and Lois Galbraith is tenacious."

Latest News from Association Barth France

By Florence Mannes, Chair, Association Barth France

For those who thought that we were on holiday last summer, here is the news :

Third Edition of the Barth France Trophy

On August 4th, the Association gathered 70 golf players at the Golf de la Bretesche in Missillac in Brittany to drive and putt for the children with Barth syndrome. The profits were donated to Barth France. The players could also make a direct donation to the Association. The sun was shining as we welcomed all the participants. Most of them had already been there for the two previous tournaments.

Thanks to this event, Barth France gathered the sum of 2500 euros. Congratulations to all the winners and many thanks to Mr. Gicquiaud, the director, for his loyalty to our cause. See you all on August 3, 2014 for the fourth edition!

Meeting the Inner Wheel Club in Tienen-Tirlemont

On September 9th, at the initiative of Brigitte Van Vlasselaer, the Inner Wheel Club of Tienen-Tirlemont in Belgium hosted a presentation on Barth syndrome. This presentation was made by Valérie, Jules's mother and by Philippe, Raphaël's father. The aim of this meeting was to talk about the syndrome through people's experiences with a presentation made by parents for the mothers that were present. The Inner Wheel Club made a 1000 euro donation to Barth France. Thanks to them!



Barth France Team at the Bruxelles Half-Marathon. (Photo courtesy of Barth France 2013)

IronBask in Saint Jean de Luz

Twenty-four triathletes from Barth France swam, rode, and ran for the Association on September 14th. The bravest chose the Ironman distances, others the Half-Ironman, and the less experienced went for an Olympic distance or a sprint. All wore the colors of Barth France with great pleasure. For more details, see Yohan and Nicola's article on page 29. The triathletes of Barth France gathered the sum of 25000 euros through this contest. Champions!

Seminary: French and English doctors at the Hôpital Necker

On September 27th, thanks to the financial support of Barth France, we organized a meeting with French and English doctors to discuss medical practice in order to create a multidisciplinary clinic in France like the one in Bristol (UK). (See Dr. Jean Donadieu's article below.)

Alcoa supports Barth France for the Paris-Versailles Run

Four Alcoa employees and two Barth France members participated in the traditional run from the Eiffel Tower in Paris to the Chateau of Versailles (17 km) on September 29th. Alcoa donated 2500 euros to Barth France for participating in this race.

Meeting with the Belgian Heart League of Cardiology

On October 7th, Barth France was received by the president of the Belgian League of Cardiology, Dr. Freddy van de Casseye. The goal of this meeting was to introduce the Barth France Association and the Barth Syndrome Foundation in order to create a network and reference centers for Belgium. We are working on how to start a collaboration. To be continued!

Save the Date: Poker Tournament Barth France, November 23, 2013

On November 23rd, in association with the Partouche casinos, we are organizing the second Barth France poker tournament. See you at the Mannes garage, 36 rue Francois Mitterrand, Ivry-sur-Seine. Registration: Philippe@barthfrance.com

New Data Published and Collaboration Viewed as Key

By Dr. Jean Donadieu, Service d'Hemato-Oncologie Pédiatrique, Hopital Trousseau, Paris, France

Thanks to Barth France, a parent association, a network of physicians has started to operate in France.

Barth syndrome (BTHS) is a multi-system disease, and so hematologists, cardiologists, and metabolic specialists all contribute to the care of patients with Barth syndrome (BTHS). It is difficult to understand the different faces of the disease and to have a fair appraisal of patients' situations without these

different perspectives. Diagnosis and care of patients both depend on this global approach. In addition, due to the low incidence of BTHS, it is almost impossible to find a center which has seen more than five cases in the last ten years. Therefore, clinical experience is rare and a registry, like that of the Barth Syndrome Foundation (BSF), is crucial to obtain a minimal level of awareness and evidence about BTHS.

(Cont'd on page 28)

New Data Published and Collaboration Viewed as Key

(Cont'd from page 27)

In France, there was only a Neutropenia Registry for Barth syndrome. So, in the fall of 2011, Barth France contacted the French Severe Chronic Neutropenia Certified Patient Registry and offered a grant to launch a study into the incidence and management of BTSH patients. Thanks to the support of Barth France, a young physician, Charlotte Rigaud, was appointed to gather data about BTSH in France. She did her data collection during the summer of 2012. Data collection is a "physical" task, and also a diplomatic task, as it is absolutely necessary to convince a huge number of physicians to share private and privileged information about their patients. In the end, after contacts with more than 100 physicians and more than 300 phone calls and emails and about 20 visits to different centers in France, information about 22 cases of BTSH was collected. This work offers a snapshot of the current practice regarding BTSH patients in France, along with their outcomes. A comprehensive report was published in the Orphanet Journal of Rare Diseases¹. (Editor's note: Please see the citation for this natural history report at the end of this newsletter article. For access to the full text, find the article in the Barth Syndrome section of the bibliography at <http://www.barthsyndrome.org> (Home >> Bibliography) and click on the link.

After this report was completed, our next step was to develop a network in France and to try to answer several questions that arose from this analysis: How to educate physicians to better recognize the disease? How to ascertain in the shortest time possible the diagnosis of BTSH? As the management of cardiac function is a key issue, what follow-up and what guidelines are needed for cardiac complications? Because our survey showed many deaths, associated with "common" viral/seasonal infections and heart failure early in life, how can these common viral infections in young patients be prevented? Lastly, how should the neutropenia be managed?

In order to develop our approach, we organized a seminar with the Bristol team, which has longstanding experience with BTSH in UK. This meeting was held on September 27th in Paris.

Organization of care

The first characteristic of the English network is its centralization around a single center: the Bristol team coordinated by Dr. Colin Steward. British BTSH patients visit the center at least once a year. In the meantime and for new patients, phone contacts between the Bristol center and patients or patients' doctors are organized. Within the Bristol team, all relevant skills are gathered: cardiologist, hematologist, laboratory biochemical, geneticist, nutritionist, and neurologist specializing in neuromuscular diseases.

In contrast, the French network is much more fragmented: It is based on four distinct reference centers (none of them dedicated to BTSH): the reference center for congenital cardiology at Necker-Enfants Malades Hospital, the reference center of mitochondrial diseases at Necker, the reference center for metabolic diseases, and the reference center for hereditary immunodeficiencies. The French Severe Chronic Neutropenia Certified Patient Registry also contributes to the collection of

data. Furthermore, the genetics laboratory and biochemistry laboratory at Necker provide biological expertise. Beyond reference centers, patients are followed in their diagnostic centers and not just at one site. The two approaches are complementary, but, from the French point of view, there would be a huge benefit to move toward a more centralized approach, given the rarity and complexity of this disease.

One option would be to organize "Barth" days in a single place, maybe twice a year, in order to offer multi-disciplinary consultations. Such "medical days" could be followed by social or cultural meetings for patients. This needs to be set up!

Diagnostic and treatment guidelines

The circumstances leading to the diagnosis of BTSH are quite stereotypical: the most common circumstance is heart failure in a boy, aged less than one year, with fever and neutropenia. But many other circumstances may lead to the diagnosis of this disease — for example, sudden death in a boy with fever, especially if there is a cardiomyopathy at autopsy. It appears especially critical to develop criteria of diagnosis in the ICU, as that probably is a first contact point. Once the diagnosis is suspected, it should be confirmed in the shortest possible time. The typical BTSH heart ultrasound features are left ventricular dysfunction due to left ventricular dilatation, non-compaction of the left ventricle, or hypertrophic cardiomyopathy. There is a consensus that 3-methylglutaconic acidemia (3MGC) is not a constant feature in BTSH but is very evocative. Finally, the profile of cardiolipin in BTSH is very peculiar. This is a quantitative and not qualitative profile and can be studied either on fibroblasts or on blood cells, according to the sensitivity of the technical process. In France, the available spectrometer requires a fibroblast culture, while in England one of the available devices allows the study to be done on leukocytes. If a typical spectrum of cardiolipin profile is observed, a genetic study of the *TAZ* gene formally confirms the diagnosis. There is consensus on this approach, but we would like to explore the possibility of performing the test on blood and not just on fibroblasts in France in the future.

There has also been discussion about the pathophysiology of BTSH, as it is a key issue to drive targeted therapy and not only symptomatic care. However, many issues remain completely open. A critical point is the metabolic management. It is imperative to consider the amino acids' chromatography profile which is peculiar in BTSH (with low arginine levels among other differences).

This result makes sense in the logic of Krebs cycle disruption and suggests that arginine supplementation could bypass the metabolic consequences of the genetic abnormality. But this needs to be balanced. There are many detailed questions to consider in this. So far, however, no conclusion can be reached due to lack of evidence, but we will try to create some guidelines.

Infection prophylaxis also is a critical issue in Barth syndrome. We observed in the French survey, that there are relatively few infections related to neutropenia. In contrast, however, we saw a significant number of "possibly viral" infections in these children

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in the first two to three years of life. These "ordinary" infections can have dramatic consequences leading to myocardial function failure. This led us to discuss the prophylaxis of infections with three existing possibilities: Long-term antibiotic prophylaxis (for example daily Cotrimoxazole), long-term G-CSF and IV immunoglobulins. Again, there are many details to discuss and work out.

In the end, with the help of the exchanges with the British group, we in France are now in the process of drawing up guidelines, for Barth syndrome and reshaping our organization at a national level!

Reference: Rigaud C, Lebre AS, Touraine R, et al. [Natural history of Barth syndrome: a national cohort study of 22 patients](#). Orphanet J Rare Dis. 2013 May 8;8:70. (Open Access)

A New Motivation — Proud of Sharing BSF Values

"We are walking in the dark, united and strong. The sun hasn't reached the horizon yet. We are alone. Faces are tensed. Eyes are focused. Hearts beat in unison, following the rhythm of the waves breaking on the shore. The sand is cold. Some purple lights on the sky are drawing the sea. It is dark. The sea breeze won't take us away from here. We are united and strong. It's 7 am, I'm in Saint-Jean de Luz, West Coast, France, and I don't want to be anywhere else. My place is here. I feel the difficulty. I understand. This feeling of difficulty-- people suffering from Barth syndrome face it everyday. Today, I'll take up the challenge. Soon, perhaps, I'll be an Ironman. But today I understand. I want to support Raphaël. I want to support his family. I want the research into the syndrome to make progress. We are walking in the dark, united and strong." ~ Nicolas Mahieu



By Nicolas Mahieu

IronBask Starting Line, Saint Jean de Luz ~ Saturday, September 14, 2013

I joined Barth France last year. I didn't know about Barth syndrome before finding the Barth France website. I was looking for an association to support while participating in the triathlon. Because I was curious to learn more about the syndrome, I decided to join the Barth France Team. We try to raise funds in order to support the medical research by showing and sharing the Barth Syndrome Foundation's (BSF) image. I met Raphaël, a 4-year old boy who has Barth syndrome, his brothers, and his family. I realized how united, strong, and determined this family was fighting for a better future for Raphaël. Now, practicing the sport does make sense.

Barth France triathletes at the Ironman Saint Jean de Luz. (Photo courtesy of Barth France 2013)

Finishing an Ironman has always been a dream, crossing the finish line, surpassing myself. Something almost impossible. Thanks to Barth France Team, I became more confident and I realized that dream on September 14th in Saint-Jean de Luz. During our training period, we helped each other surpassing ourselves. I felt stronger and encouraged. I think Barth Syndrome Foundation members have values that exactly fit suffering peoples' needs. More than realizing a personal challenge, I joined an altruistic way of thinking, and I made friends. I look forward to participating in another challenge, feeling that people talk more about the syndrome. I know that medical research will continue to move forward.

Today, I know that I have grown up myself. I'm practicing sport in a pleasant environment. I found a new motivation. I hope that every trip and challenge we organize will contribute to the cause. Today, I'm proud of sharing BSF's values. We are going forward together, hoping that one day, we'll reach our goal!

A New Barth Story That Begins...

By Yohan Villeudieu

It was in October 2012 and I was looking for a club in Paris with training hours that suit me. I randomly arrived on a triathlon website that mentioned Barth France. It took me only a few minutes to contact by e-mail Philippe, Raphaël's father, whose son has Barth syndrome.

This is how my story with Barth France started. Two weeks later, I was meeting the Mannes family during a visit of my firehouse in Paris. I can't remember if it was Raphaël or me that was the most impressed. He is 4 years old, the same age as my son. He had his eyes wide open and filled with joy when he went on a fire truck, and I was so happy to meet a little hero with an unforgettable smile, who fights an unknown disease everyday. This is what convinced me to

join the Saint Jean de Luz Ironman and to wear Barth France's colors to represent the disease and eventually eradicate it!

This year was quite busy for all of the Barth France members and I must say we do not like simple: How to gather everyone for an Ironman when the members are entrepreneurs, running four bakeries, head of communication ... without neglecting your family. It is quite simple: You just tell yourself that you are doing this for these kids who fight everyday and it works.

The Ironman is over, and we all did it entirely. Now we must go ahead and remember that even when things seem tough, like the disease, we can go through. Join US!!! Let's make a difference!

Top Ten Reasons to Give BSF Your Email Address

1. You'll hear news quickly. 2. It's free. 3. It's easy for both of us. 4. We like to say thank you. 5. We'll save on postage and can put those \$ toward our programs. 6. If you move, we can still stay in touch. 7. You can forward BSF messages to your friends and help us increase awareness. 8. We promise not to sell your name or to send you too many messages. 9. You can unsubscribe easily at any time. 10. Because you care about our mission.

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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.



Christopher (age 5).
(Photo courtesy of BSF 2013)

Thank You for Making a Difference!

Your generous donation will give hope to our community... Hope to boys and young men, like Christopher, who simply want to be healthy and strong.

From all of us here at BSF, thank you. Thank you for allowing us to persistently work toward our vision...

a world in which Barth syndrome no longer causes suffering or loss of life.

BSF Wins Spot on 2013 Top-Rated List

BSF once again won a spot on the GreatNonprofit's 2013 Top-Rated List. BSF is one of the first nonprofits to be honored this year and we are now listed as a winner on Great Nonprofit's leaderboard. Thank you to all those who submitted a review in support of BSF!



Power of Kindness



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

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Jeremiah (age 15) and Henry (age 5). 2012



Will (age 27) and Bly (age 9). 2012

(Photos courtesy of BSF)

Power of Kindness

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

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

~ Affected Family, Great NonProfit Review, 2013

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William (age 5) and Benjamin (age 9). 2012



Raphaël (age 4) and John (age 31). 2012

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Joe (age 5). 2013



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(Photos taken for BStrust by Samantha Holmes)

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(L-R) Robert, Adam, Chris, and Travis at BSFCa's 2013 Golf Tournament.



(L-R) Adam, Ryan, and Travis.



(L-R) Adam, Ryan, Robert, and Travis.

Editor's note: Robert (age 27), Adam (age 24), Ryan (age 22) and Travis (age 18). (Photos courtesy of BSFCa 2013)



Barth Syndrome Foundation

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Barth syndrome (BTHS; OMIM #302060)

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

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

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



Henry (age 5) ²⁰¹³

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