



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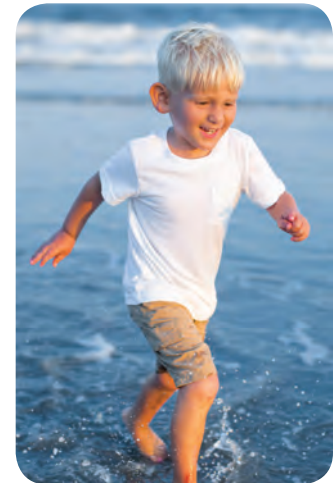
Saving lives through education, advances in treatment, and finding a cure for Barth syndrome.

Will We Be Doing Enough?

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

“We few, we happy few, we band of brothers... ” We are now entering a period when we need a Barth band of brothers, just like King Henry V.

In my graduate student days there was a poster that hung in the laboratory of a fellow graduate student that contained the quote that suggested the title of this essay. I guess it was meant to inspire us to work harder, faster, and better — sort of a mantra for the graduate student program. The actual quote, “You have not done enough, you have never done enough, as long as there is still something to which you can contribute,” is from the second UN Secretary-General, Dag Hammarskjöld, who was probably referring to people working for world peace. Working for world peace is a favorite answer of many prospective candidates for office, or beauty contestants. Nevertheless, for us seeking to find a treatment for Barth syndrome, it can also apply.



Connor
(Photo courtesy of Connor's family)

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The Barth syndrome community is made up of individuals with the disease, their family and friends, their physicians, and all those researchers and BSF members working towards that common vision of a world where Barth syndrome no longer causes suffering or loss of life. We are now at a time in history where something can be done to help bring this about. Over a half dozen therapeutic ideas are in various stages of development right now, and these ideas will soon

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Major Milestone Reached by BSF: ICD Code E78.71 for Barth Syndrome

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

Sometimes the simplest of questions are the most difficult to answer. What are the incidence (newly diagnosed over span of time), prevalence (total population), morbidity (measure of illness) and mortality (measure of death) of Barth syndrome (BTHS)? It's complicated. We can provide de-identified metrics about those who have contacted us and enrolled in our Barth Syndrome Registry, but until recently that was the extent of our metrics. We had limited data to measure disease severity for those who had Barth syndrome on a global level. Nor did we know the number of deaths caused by Barth syndrome or how many people had been diagnosed with Barth syndrome worldwide.

From very early on, we at the Barth Syndrome Foundation (BSF) came to agree that ultimately one of the best ways to gain information about that unknown group was through the creation of an International Classification of Disease (ICD) code specifically for our disorder. Since BSF was established, it became one of our goals to have a distinct ICD code for Barth syndrome. While not infallible, an ICD code would provide a way to know just how many people were diagnosed with Barth syndrome.

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We Are Family... I Got All My Brothers With Me

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

Do you remember that song in the 70's by Sister Sledge, "We Are Family"? That song was running through my head quite a bit as we recently hosted a volunteer workshop in South Carolina with the staff, the Board of Directors, and some of our affected families.

The goal of the workshop was to find ways to further engage the wider community through increased volunteer efforts and reinforcement of our mission. On the first night, we asked attendees to share what being a part the Foundation meant to them. Jack, age 22, shared that while BSF stands for Barth Syndrome Foundation, of course, for him, what it really stands for is **Barth Syndrome Family**. Jack explained that his "Barth Family" is where he feels normal and loved, just like with his own relatives.

Throughout the course of our three-day retreat, what Jack said about family stuck with me. In our lakeside cabins, we shared meals and talked about the early days at BSF. The organization was built through the hard work of volunteers with blood, sweat, and many tears. Much of the grassroots culture still remains present, yet the organization has grown so much in these past 15 years.

Unlike those early years, we now have paid staff in addition to our dedicated volunteers, to carry out the work needed to find treatments and a cure. We are closer than ever before. This is thanks to the efforts of those who started the Foundation and to the generosity of our donors. You are all part of our Barth family.

During our time together discussing our mission, I witnessed a passion like I've never seen. Emotions were high. A few brave souls admitted to feeling unsure of whether or not their contributions mattered. We all listened to each other. And you know what happened? Compassion, empathy, and love filled the room. In the end, everyone realized we had each other's backs... just like true family.

(Cont'd on page 3)



(L-R) Shelley Bowen & Lindsay Groff



(L-R) Bill Zeller, Steve McCurdy,
Florence Mannes & Matt Toth



(L-R) Susan McCormack & Stacey Woodward



Matt Toth presenting upcoming scientific projects

We Are Family... I Got All My Brothers With Me

(Cont'd from page 2)

We may approach things from different perspectives at times, and we have certainly made mistakes. But, we are all here because we care about someone affected by Barth syndrome. In the end, it's what unites us that matters most. Like all families, there are laughs and tears. There are celebrations and heartaches. Through it all, we find a way to show kindness, understanding, and grace.

All of those things were in full effect when we all came together with a common focus this past weekend. I'm excited to take what I learned from the workshop into my daily work with BSF. I'm very happy to share that we achieved our goal of bringing people together with a common mission: *saving lives through education, advances in treatment, and finding a cure for Barth syndrome*. Thank you for being part of our family.



(Front Row L-R) Anne Zeller, Florence Mannes, Cathy Ritter, Stacey Woodward, Susan Osnos, Steve McCurdy, Sheila Mann, Michelle Florez, Kevin Woodward (missing from photo is Bill Zeller)

(Middle Row L-R) Nicole Derusha-Mackey, Lindsay Groff, Lynda Sedefian, Susan McCormack, Jack Higgins, Shelley Bowen

(Back Row L-R) B.J. Develle, John Wilkins, Matt Toth, Marc Sernel (All photos courtesy of BSF²⁰¹⁵)

In keeping with BSF's mission, the primary goals of the workshop were:

- To gain a deeper understanding of potential treatments that are on the horizon and their prospective impact on our community.
- To develop plans to improve BSF's fundraising, marketing, communications, and family services to better engage and serve our community as we move forward.

The participants spent over 480 total person-hours brainstorming, contemplating, discussing, and debating these issues. We're enormously excited about the ideas that were generated and the momentum we'll generate going forward.

Several new committees were established that will be organized in the coming weeks. There will be calls for additional volunteers to perform various duties – please consider helping us. BSF needs you!

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Thanksgiving Thoughts

By Marc Sernel, Chairman, Barth Syndrome Foundation

Thanksgiving in the U.S. is a time for family and reflection, pausing from our busy daily lives to think about the things for which we are thankful. For my family, it's also historically been a week when we head down to warmer weather in Florida to enjoy a vacation, which we did again this year. We have had to cancel our Thanksgiving trip plans twice — once when my twin boys were five months old and then again when they were in first grade — each time because my son Ryan was facing an acute health problem. Because the timing of my son's prior health scares have always seemed to coincide with the run-up to Thanksgiving, this is a time of year when I instinctively reflect on Barth syndrome, the Barth Syndrome Foundation (BSF), and giving thanks.

My family has been dealing with Barth syndrome for a decade (wow -- even after typing that it's still hard to believe). It is so much a part of our "normal" every-day life today that it is nearly impossible to remember the "pre-Barth" days. Thrice-daily medication and frequent trips to the cardiologist, among many other sacrifices, are just part of our normal routine. Having children changes a person's life in many ways, placing restrictions on what you are able to do but offering intangible rewards that usually far outweigh the sacrifices. Having a Barth child means facing many more and greater challenges — challenges you didn't think you were signing up for when deciding to have children — but those challenges also bring a greater perspective and appreciation of the small things and what really matters. I for one am a different and better person because I have a child with Barth syndrome.

All of us who are part of the Barth Syndrome family know that while we have been dealt challenges that are greater than most, we also know that we have a lot to be thankful for. If my son was going to be affected by a very rare, life-threatening disorder, I am very thankful that it turned out to be Barth syndrome so that we had this amazing organization to help us through the journey. It

is an oft-repeated saying but it is so true that it bears repeating: "None of us would have ever chosen to be a part of this group, but since fate has dealt us this hand we sure are glad that BSF is there for us."

I am eternally thankful for all of the various contributors that make BSF what it is. I am thankful for our amazing BSF staff members, whose extreme competence in their jobs is surpassed many times over by their passion and extra-mile commitment for the health and well-being of those affected by Barth syndrome. I am thankful for those whom I serve with and have come before me on the BSF Board of Directors. All of them are amazing people who have offered their precious time and many talents to build and grow our organization. I am thankful to all of our families; those who started this journey before we did, blazing the trail and offering life-altering and -saving advice to my family, and those who walk with us and follow in our footsteps, committing themselves to the cause and helping us collectively move toward our goals. I am thankful for the many doctors and researchers who have worked tirelessly over the years to learn as much as we have about this terrible disease, and now help the boys live better and longer and have put potential treatments nearly within our grasp. I am thankful for the incredible generosity of our volunteers and donors, without whose time and treasure much of what we do would not be possible. In short, if you are reading this, I am thankful for you and everything you've done to help this organization and our boys.

My family cherishes our time in the Florida November sun before heading back north for the cold Chicago winters, just as we try to cherish the little things and small moments that we enjoy with our family and friends. I am thankful, and we should all be thankful, that we have each other in this amazing Barth Family. Happy holidays to everyone and thank you for your support.

Meet the Barth Syndrome Foundation's Board of Directors

(Photos courtesy of BSF)



Marc Sernel



David Axelrod



Matt Blumenthal



Randy Buddemeyer



Florence Mannes



Susan McCormack



Susan Osnos



Cathy Ritter



John Wilkins



Kevin Woodward



Steve McCurdy



Lindsay Groff

Will We Be Doing Enough?

“Over a half dozen therapeutic ideas are in various stages of development right now, and these ideas will soon need Barth guys to volunteer for treatment trials/studies. Will they? We are “building it,” but will they come?”

(Cont'd from page 1)

need Barth guys to volunteer for treatment trials/studies. Will they? We are “building it,” but will they come? It is a critical question.

I have just come from a conference where many rare disease groups were represented. The meeting rejoiced in the realization that patients are becoming more and more important in how drugs are approved and even discovered. Apparently, the 21st century is going to be the patient-centric century. Drug makers and physicians have to include the patient's input into the next generation of drugs and medical devices. Unlike our ultra-rare disease, many other rare disease groups suffer a dire prognosis for the future of their affected members. Clinical science can predict that many individuals with these diseases will have a bad outcome. In our disease, there is no such certainty, but we do know that today no one who has Barth syndrome gets better. It has also been true that some of our guys get very sick and even die, very often without much warning.

We need to change that reality as fast as possible. But even after all the research and researchers do their job, in the end progress is really up to the current group of Barth guys to volunteer to test these therapies. There is no one else who can do that job. No one likes to take risks if there is a better or more certain alternative, but we will have no new options if new possibilities are not tested, and we will have to continue doing the best we can with what is available now. But what is available now is not acceptable.

Volunteering for a treatment study is a personal/private decision. There are no guidelines from the volunteer's perspective, but there may be some limitations from the treatment trial, like age, current health status, etc. Those trial limitations are imposed to make sure that everyone who volunteers counts and that their sacrifices to participate mean something. The real questions Barth guys have to ask themselves are: If not me, then who? If not now, then when? If not this treatment trial, then which one?

One of the most popular BSF videos is one where Ben is introducing one of the SciMed sessions of the 2010 BSF Conference. Ben said to all the scientists and doctors there, “. . . please give us treatments to try, and then if they don't work we'll try again.” It cannot be said any better!

To all the Barth guys, young and old, please keep yourself informed. If you need help to understand what is going on, ask someone. When the time comes that volunteers are needed, consider the call seriously. You are, by definition, few. Shakespeare penned perhaps the best prose about how a few people can change the world: “We few, we happy few, we band of brothers... .” We are now entering a period when we need a Barth band of brothers, just like King Henry V.

Editor's Note: Please visit the Barth Syndrome Foundation's website to learn about treatment studies currently underway for Barth syndrome.



Dr. Matt Toth introduces Ben at BSF's 2010 Conference
(Photo courtesy of BSF²⁰¹⁰)

(Photos courtesy of Amanda Clark²⁰¹⁴)



Emily & Luke (age 3)



Levi (age 3)



Jack (age 5)



Wyatt (age 8)



Noah (age 14)

Major Milestone Reached by BSF: ICD Code E78.71 for Barth Syndrome

(Cont'd from page 1)

The compliance deadline of the 43rd World Health Assembly by member states of the World Health Organization (WHO) was October 1, 2015. In this 10th version of ICD codes Barth syndrome was assigned a specific ICD code (E78.71).

We have earned the reputation as the authority of source about Barth syndrome. Therefore, it is reasonable to receive inquiries about incidence, prevalence, morbidity, and mortality. With this specific code we now have a tool to measure Barth syndrome health statistics in the broader population around the globe, in addition to the more detailed data we collect through the Barth Syndrome Registry.

This is a major accomplishment! We have strived to have a distinct ICD code since we were first formed in 2000. It wasn't possible to simply add a number for Barth syndrome. It was a huge, multi-country undertaking with complicated international bureaucratic processes. It took us 15 years and a great deal of multinational teamwork for this goal to be achieved. Go BSF and our affiliates!

Dr. Gerald Cox's New Role

By Kate McCurdy, Emerita, Scientific and Medical Advisory Board, Barth Syndrome Foundation

Gerald F. Cox, MD, PhD has been a valuable member of our Barth syndrome family since we had our very first gathering in June 2000 in Baltimore. Then, after BSF was created, he has served on our Scientific and Medical Advisory Board (SMAB) since its inception in 2001. Dr. Cox has brought an important perspective to this group as the only member who works full-time in "industry" (as Vice President of Clinical Development in the Rare Diseases Group at Genzyme Corporation, which is now part of Sanofi) in addition to holding an appointment at a teaching hospital (as an Attending Physician in Genetics at Boston Children's Hospital). With experience as both a scientist and a physician, he has made significant contributions as a grant reviewer and an advisor regarding our registry, and he also has offered good clinical advice to a number of Barth patients.

Now that we are getting ready for clinical trials, we at BSF have asked him to focus his attention on advising our Science Director, Matt Toth, on issues related to these evolving efforts, so Dr. Cox will step down now from his regular SMAB duties. We are all very grateful to him for his long service on the SMAB, and we look forward to continuing our work with him on this new and exciting front. Thank you, Gerry, for all you have done and will continue to do for the Barth Syndrome Foundation!



Dr. Gerry Cox (center) with families and physicians gather in Baltimore in 2000 for the first-ever Barth syndrome meeting
(Photo courtesy of BSF²⁰⁰⁰)

New Members Join Barth Syndrome Foundation's International Scientific and Medical Advisory Board

By Catharine Ritter, BSF SMAB Member, Ex-officio Member; Board Director, Barth Syndrome Foundation of Canada

In the 15 years since the first Barth Family Meeting in 2000 and the subsequent formation of the Barth Syndrome Foundation (BSF), the growth and accomplishments of the Foundation have been outstanding. One of the greatest accomplishments has been the development of the international Scientific and Medical Advisory Board (SMAB) which was established by Kate McCurdy, former BSF board member and now *Emerita* member of the SMAB. The SMAB's membership is made up of expert doctors, researchers, and scientists in the various areas where Barth syndrome manifests itself. They help develop new research to learn more about this disorder and into possible treatments; they review all of the research grant applications and provide advice to the Board on where to channel the precious research funds. In order to keep the SMAB fresh and dynamic while moving forward, new members with their unique ideas, perceptions, and expertise are occasionally added to strengthen and grow the SMAB. With this in mind, the BSF is pleased to announce that John Lynn Jefferies MD, MPH, FAAP, FACC and Hilary Vernon, MD, PhD, have joined the SMAB. Both Dr. Jefferies and Dr. Vernon are familiar members of the Barth syndrome community, having attended and spoken at a number of the BSF's international conferences.



Dr. John Lynn Jefferies

Dr. John Jefferies is a Professor of Pediatric Cardiology and Adult Cardiovascular Diseases and the Director of Advanced Heart Failure/Cardiomyopathy within the Heart Institute at Cincinnati Children's Hospital Medical Center. He completed his combined Pediatric and Adult Cardiology training at the Baylor College of Medicine in Houston, Texas at the Texas Children's Hospital and the Texas Heart Institute. *(Photo courtesy of Dr. Jefferies²⁰¹⁴)*

Dr. Jefferies's current research interests include heritable causes of vascular disease, novel drug therapies for advanced heart failure, novel gene discovery in cardiomyopathy, characterization and management of left ventricular noncompaction (LVNC), and early diagnosis and management of chemotherapy induced cardiotoxicity. One of Dr. Jefferies's current research projects addresses the quality of life experienced by the boys and men affected by Barth syndrome.

He is on the Editorial Board of the Texas Heart Institute Journal and is an active member of numerous professional organizations including the Heart Failure Society of America, the American College of Cardiology and the American Heart Association.



Dr. Hilary Vernon

Dr. Hilary Vernon is an Assistant Professor of Genetic Medicine at the McKusick-Nathans Institute of Genetic Medicine at Johns Hopkins University and at the Kennedy Krieger Institute (KKI) where she is the Director of the Barth Syndrome Interdisciplinary Clinic. Dr. Vernon also serves on the Maryland State Advisory Council on Hereditary and Congenital Disorders. She earned her MD and PhD at Rutgers University and completed residencies in Genetics and Pediatrics at Johns Hopkins University and a fellowship in Clinical Laboratory Biochemical Genetics at Johns Hopkins University. Dr. Vernon is board certified in Pediatrics, Clinical Genetics, and Clinical Laboratory Biochemical Genetics. Her main areas of research include understanding intermediary metabolism in Barth syndrome and in disorders of branch chain amino acid metabolism. Dr. Vernon knows many of the Barth guys well because she sees them as the metabolic expert at the Barth Syndrome Clinic that is held four times a year at KKI. She is collecting longitudinal data that will increase the understanding of this disorder and may be helpful as we enter our new phase of clinical trials. *(Photo courtesy of Dr. Vernon²⁰¹⁴)*

The Barth Syndrome Foundation Board of Directors is excited and honored to welcome Drs. Jefferies and Vernon to the SMAB. Their experience in both areas of clinical practice and research, as well as their expertise in Barth syndrome specifically, will be invaluable additions.

(Photos courtesy of Amanda Clark²⁰¹⁴)



Ben (age 11)



Jake (age 17)



Derek (age 22)



Andrew (age 26)



Joshua (age 32)

We're One Family Working Toward One Goal

By Jack (age 22), Affected Individual, New Jersey

"I was asked what is it like to have Barth syndrome? What it means to me to have Barth syndrome is that, although I was diagnosed as having a rare disease, I was also welcomed into one of the greatest families in the world. I think that the initials "BSF" do not only stand for the Barth Syndrome Foundation but, in fact, it stands for the "Barth Syndrome Family!"

On November 12-15th, the Barth Syndrome Foundation (BSF) held a workshop graciously hosted by the Bowen family in South Carolina. The idea of the workshop, in my opinion, was to update us on the progress we have made in the past year and a half since the last conference. What we learned is that there are upcoming clinical studies on possible drugs to help the affected boys and men. We also learned what we can do to help continue the great research progress that has been taking place over the last 13 years. For example, we will need more participants (affected guys) to step up and pitch in with all that will be going on. We need more than the "core" group of guys to participate in the studies and treatment trials that are or that could be going on in the near future. I also was asked what it is like to have Barth syndrome? What it means to me to have Barth syndrome is that, although I was diagnosed as having a rare disease, I was also welcomed into one of the greatest families in the world. I think that the initials "BSF" do not only stand for the Barth Syndrome Foundation but, in fact, it stands for the "Barth Syndrome Family!"



Jack (age 22)
(Photo courtesy of Amanda Clark²⁰¹⁴)

Since the Foundation was formed back in 2000, many new families have joined the "Barth Syndrome Family." I feel one of the reasons that we do not have more participants in studies and at conferences is because of fear of being an outsider. Newcomers might feel weird or outside their comfort zones asking questions about stuff that some of the rest of us have learned over the years. But in reality, there is only the "Barth Syndrome Family"! We are one big group, all looking for one goal — a cure! We are also a group of warm, welcoming, and always excited families eager to answer any questions that a new family might have in regards to this disorder. Everyone struggles with Barth syndrome — parents, siblings, affected children — and we are all trying to deal with living well, daily.

In the end, we are one big family headed for the light at the end of the tunnel which is the cure for Barth syndrome! So, to all the newcomers out there, come this summer when we host our biennial conference. I encourage you to feel free to ask away — don't hesitate to walk up to one of the families that have had past experiences and ask about anything that is on your mind. Never be afraid to ask, because we all share the same thing. And we all want the same thing..... a cure! Together, we can help that day come sooner.



(L-R) B.J. Develle, Lindsay Groff, Nicole Derusha-Mackey, Stacey Woodward & Jack dress for the 80's Flash Dance show
(Photo courtesy of BSF²⁰¹⁵)



(L-R) B.J. Develle, Steve McCurdy, Florence Mannes, Susan McCormack & Jack at the Volunteer Workshop
(Photo courtesy of BSF²⁰¹⁵)

The Value of BSF From a Grandparent's Perspective — Small but Sensational

By Anne Zeller, Grandparent of Rhys and Bryn, New Hampshire

I went to my volunteer job at the Red Rock State Park Visitor Center in Sedona, Arizona earlier than usual that February morning in 2011. We normally left the answering machine on until we opened, but something made me pick up the ringing phone and I heard the worst words a parent or grandparent can hear... "Thank God I've reached you! There's been a terrible tragedy!" Scenes of mangled autos and my family all killed raced through my mind and it took another second or two for the words, "Your grandson, Rhys, has died" to register. Confusion, disbelief, and helplessness at being 2000 miles from our children in New Hampshire with no practical way to get to them in a hurry paralyzed me. These things happened to other families, not mine!

As everyone knows who has lost a child, no matter how young or old, grief can be difficult to overcome, especially when the cause of death is unknown. When the test results revealed Rhys, at age six months, had died of complications from Barth syndrome (BTHS), a genetic disorder, and our daughter, Kate, had already given birth to Bryn, I rushed to the internet to find the answers that the doctors couldn't seem to supply. The information I found was not encouraging. It was terrifying.

When Kate and Sandt's second son, Bryn, was diagnosed with Barth syndrome, we gave mighty thanks to the doctors, nurses, and technicians at Dartmouth Hitchcock Medical Center in Lebanon, NH, for their intensive efforts to learn everything there was to know about the treatment of BTHS and to close family friends for directing us to the Barth Syndrome Foundation (BSF). At last, here was a resource where positive information was spread and through which research could be shared. The kids attended their first conference in Florida when Bryn was just six months old, and the support they found was amazing.

As a grandparent, BSF has given me a way to not only become more educated about Barth syndrome, but also to help spread awareness of this rare disease. Most of all, it gives me great happiness to see the friendships, made possible by BSF, fostered around the world between the courageous boys and their families. Although we still grieve for Rhys, thanks to BSF, we have hope for a bright future for Bryn and his "Barth brothers" and feel empowered knowing that help is only an email or phone call away.

I'm looking forward to meeting other grandparents at the 2016 Barth Conference. Together we may be small, but we are sensational! We are so grateful to BSF for all the resources they make available to the families and for funding research to find a cure!



Anne & Rhys (7/27/2010-2/10/2011)



Rhys & Sandt²⁰¹¹



Bryn (age 4) and his best friend, Teddy



Bryn enjoys riding his therapy horse while Kate looks on proudly
(Photos courtesy of Anne Zeller & Kate)

A Few Caring People Can Change the World

By Sandra Stevens, Proud Volunteer, Barth Syndrome Foundation

“Never believe that a few caring people can’t change the world. For, indeed, that’s all who ever have.” ~ Margaret Mead

Sometimes we’re lucky enough to find a job that produces more than just a paycheck. During the time I was employed by the Barth Syndrome Foundation (BSF), I got to know a wonderful group of truly inspiring people, tirelessly dedicated to ridding the world of Barth syndrome and helping those who suffer from this terrible disease. Not only have I seen families of affected boys and men overcome their own challenges to raise awareness and funds for BSF, but friends and neighbors have also been busy, and the simple fact is that BSF couldn’t do any of the work they do without this support. Your donations and fundraising efforts are the life-blood of the organization.

Like you, I’ve found that it’s impossible to spend time with these amazing young boys and men and not want to do something, however small, to make their future brighter. With every dollar you donate, or minute you spend supporting BSF, you are helping to do just that. There are just a few weeks left before the end of the year, so let’s make sure 2015 beats all the records!

So much has already been achieved this year. Starting with the Happy Heart Walk in February and other events I listed in the June newsletter, everyone who has been touched in some way by Barth syndrome has continued to come up with great ideas to raise awareness and funds. Your gifts are precious to BSF, and it’s my pleasure to list the latest group of heroes. You, and all those who support you, are all winners in my book!

Charity Bingo for Barth Syndrome – June 11, 2015 (Kansas City, OH)

Bryan, father of Abram, organized this now annual fixture in a new venue this year, and it turned out to be bigger and better than ever. Thank you, Brian, and all those who contributed to BSF. *(Photo courtesy of Bryan²⁰¹⁵)*



Abram (age 8) and Bryan at the Charity Bingo

Community Yard Sale – June 13, 2015 (Sewell, NJ)

Lindsay Groff, together with other neighbors, held her 4th annual yard sale, with a portion of the funds raised going to support BSF. Proof that even something you no longer need may be precious to BSF. *(Photo courtesy of Lindsay Groff²⁰¹⁵)*



Community Yard Sale

Breaking Barth 2015 – July 25, 2015 (Chapel Hill, NC)

Our Breaking Barth hero of 2014, Michael Neece, along with Master Neill and many dedicated students of Neill's Taekwondo and Fitness, broke over 1,000 boards and raised funds to support Barth syndrome this year! In 2014, Michael broke his hand while preparing for the first Breaking Barth event. We’re very pleased that this year, the team chose to break only the previous record, and no bones at all! *(Photo courtesy of Michael Neece²⁰¹⁵)*



Michael Neece prepares to break boards for Barth

City2Surf 2015 – August 9, 2015 (Sydney, Australia)

In Sydney, Australia, Eli’s mom, Elissa, and her friends successfully completed the City2Surf race. After finishing the 14km run — the world’s largest — Elissa herself said, “We had an amazing day! Everyone in “Team Eli” completed the run in high spirits. We smashed our goal, and we appreciate everyone’s kind words of encouragement and generous donations. We are also extremely grateful for our amazing friends who ran in ‘Team Eli’ this year. We couldn’t do it without their love and support!” *(Photo courtesy of Elissa²⁰¹⁵)*



Elissa and Eli

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A Few Caring People Can Change the World

(Cont'd from page 10)

Ludlow River Pirate Day – September 19, 2015 (Ludlow, KY)

NKY Fencing Academy (NKFA), Second Sight Spirits and Wynner's Cup Café partnered together to create the Ludlow River Pirate Day! It was LEGENDARY, with approximately 45 fencers! Outside activities were phenomenal, despite getting foiled(!) by rain. Thanks to Ned, NKY Fencing Academy, and all those who supported this event and contributed their pieces of eight to the treasure chest. Oooaaargh. *(Photo courtesy of Ned²⁰¹⁵)*



Ned (center) & his fencers at Ludlow River Pirate Day

Devin's League of Superheroes – November 6, 2015 (Flint, MI)

To celebrate the 11th year since Devin's heart transplant surgery, Nicole, Sarah and their friends and family hosted a superhero-themed night of bowling, food and fun. Devin's League of Superheroes was formed just a year ago and already it's an awesome fundraising machine! I can't wait to see what they come up with in 2016! *(Photo courtesy of Nicole²⁰¹⁵)*



Devin at the League of Super Heroes fundraiser

Team Will

The entire Barth community suffered a terrible blow with the loss of Will McCurdy last year. That devastating news affected everyone very deeply and no one would really have blamed the members of Team Will if they felt so discouraged that they hung up their running shoes for good. Although it was through meeting Will that they were first inspired to form Team Will, it was also through Will that they learned about the other affected boys and men. It's the daily battle of those still living with this disease that keeps them running, cycling and swimming hundreds of miles between them. Now a whole new generation of Team Will members are following their parents' lead. Here are just a few of the events they've taken part in over the last few months.



Stefan Tunguz at the IronMan Lake Tahoe

IronMan Lake Tahoe – September 20, 2015 (Lake Tahoe, CA)

Stefan Tunguz is now an IronMan veteran, and we all still remember his amazing achievement last year when he finished the most grueling of them all in Kona, Hawaii. Just because he's completed so many IronMan courses, let's not forget that we are talking about one of the hardest physical tests of endurance that exists. Also, Stefan didn't attempt his first one until he was already 50 years old. *(Photo courtesy of Stefan Tunguz²⁰¹⁵)*



(L-R) Chris Fairchild, Heather Segal, Laura Azar, Gary Rodbell & Stefan Tunguz at Westchester Triathlon

Westchester Triathlon – September 25, 2015 (Rye, NY)

On September 27th, Heather Segal and Laura Azar took part in the annual Westchester Triathlon. On that day, they were cheered on by fellow Team Will members Gary Rodbell, Stefan Tunguz and Laura's husband, Chris, who celebrated with them afterwards. See Heather's article on pages 12-13. *(Photo courtesy of Heather Segal²⁰¹⁵)*

IronMan Maryland – October 17, 2015 (Cambridge, MD) Philadelphia Marathon – November 21, 2015 (Philadelphia, PA)

Jaime Jofre was briefly upstaged by Hurricane Joachim, but once the IronMan Maryland was rescheduled, he was able to get a little closer to his goal to travel 250.2 miles for Team Will and BSF in 2015. DONE! Jaime just completed the Philadelphia Marathon on November 21st, nailing this incredible achievement. Jaime also completed the NYC Half Marathon on March 15th and the IronMan 70.3 in Mont Tremblant, Quebec on June 21st. I wonder what he has planned for 2016? *(Photo courtesy of Jaime Jofre²⁰¹⁵)*



Jaime Jofre at the Philadelphia Marathon

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A Few Caring People Can Change the World

(Cont'd from page 11)

Marine Corps Marathon — October 25, 2015 (Arlington, VA)

Julia Rodbell is the latest member of the Rodbell family to join Team Will's ranks, following in the footsteps of her father, Gary, quite literally. I think the legacy of one brave young man is in very safe hands. Thank you, Julia. *(Photos courtesy of Julia Rodbell²⁰¹⁵)*

#GivingTuesday — December 1, 2015

We did it. In ONE DAY, BSF raised \$35,217. We did it together, and we did it with LOVE. Thank you all for pushing hard to make a difference in the lives of these boys and men affected by this horrible disorder. That's thanks to you, our beautiful BSF family. Imagine the possibilities in what we'll tackle next!



Michael Mancino & Julia Rodbell
at the Marine Corps Marathon



Collette, Julia & Gary Rodbell at the Marine Corps Marathon

This Year, It's Different

By Heather Segal, Member of Team Will, Larchmont, NY

“A hero is an ordinary individual who finds the strength to persevere and endure in spite of overwhelming obstacles” ~ Christopher Reeve

To all the Barth boys and men out there: **YOU** are heroes!

While training for this year's Westchester triathlon, I kept feeling it: this year is different. For the past nine years I've competed in triathlons as part of Team Will to raise money to support the Barth Syndrome Foundation. This year was different because instead of raising funds in honor of Will McCurdy (after whom Team Will was named), I was now raising funds in memory of Will.

Those of you who knew Will, or who know anyone with Barth syndrome, understand that for the Barth boys and men, every day can feel like a triathlon. A triathlon for which there are no medals.

As I hopped onto my bike to train for this year's Westchester triathlon, a memory came flooding back to me. I vividly recalled arriving at the hotel in Tempe, Arizona back in 2009. I was there to compete in my first Ironman triathlon. A proud member of Team Will, I got out of the taxi excited to check in and meet up with my Team Will friends. As I looked around the hotel lobby, I saw many other Ironman competitors there. Unlike me, they boasted top-of-the-line bikes, fancy helmets, and perfect bodies to accompany their gear. I overheard these triathletes discussing their training times, heart monitors, splits, and race gear. I remember feeling a strong sense of imposter syndrome – what am I doing here among all these real triathletes?

(Cont'd on page 13)

This Year, It's Different

(Cont'd from page 12)

But then it struck me. Most triathletes, and likely all those in the lobby that day, strive for a PR, or Personal Record, each time they race. Impressively, they aim to swim, bike, and run faster than they have ever done before. I also strive for a PR, but it's a different one. I define my PR by fundraising dollars, not triathlon speed. Each year, I try my best to raise more money for BSF than the year before.

Now, let's get back to this year, and to the day of the September 27th Westchester triathlon. When I woke up that morning, I continued to feel that this year was different. The race started, and I felt surprisingly strong despite having trained less than usual for this race. Toward the end of the run, though, I felt myself getting tired, and thought about whether to walk a little. No big deal, right? Just then I felt a gentle push — perhaps wind? — from behind my back, and looked behind me. There was no one there. I thought of Will and imagined that maybe he was gently guiding me toward the finish line.

At the Westchester triathlon, Team Will triathletes always benefit from the support of awesome Team Will fans who cheer like crazy as we triathletes approach the finish line. This year, though, only a handful of Team Will members participated in this race because one had just completed an Ironman race the prior weekend, and others would be competing in Ironman races the following weekend. I expected I wouldn't see any Team Will fans at this year's finish line. For the first time, I'd have to finish this race alone.

Suddenly I felt — or was it heard? — Will's voice gently whispering, "Don't worry about it. I'll be there with you at the finish line." The memory of Will's courage gave me strength, and I ran that last mile with renewed energy. As I approached the finish line, to my delight, I saw a group of loyal Team Will spectators smiling and cheering as loudly as ever. Their shouts of "Go, Heather! You got this!" gave me a final burst of energy, and I sprinted to the finish line, a big smile on my face.

Thanks to the generosity of friends and family, I raised over \$11,000 for the Barth Syndrome Foundation this year, and achieved a new personal record. *Will, we did it!*



Heather Segal



Team Will in 2012
(with Will McCurdy in the driver's seat)



Heather Segal & Laura Azar



Team Will triathletes
after a Westchester Triathlon race
(Photos courtesy of Heather Segal)

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 120 articles have been published on BTHS research conducted with the support of BSF and/or BSF affiliate funding (denoted below with *) and/or acknowledge biological samples and/or information from Barth families, the Barth Syndrome Registry and Repository, and/or BSF affiliates (denoted below with Δ). Listed below are articles relevant to BTHS that have been added to BSF's library since the last issue of the Barth Syndrome Journal. To view the complete bibliography on BTHS, please visit www.barthsyndrome.org.

1. Denning C, Borgdorff V, Crutchley J, Firth KSA, George V, Kalra S, Kondrashov A, Hoang MD, Mosqueira D, Patel A, Prodanov L, Rajamohan D, Skarnes WC, Smith JGW, Young LE. **Cardiomyocytes from human pluripotent stem cells: From laboratory curiosity to industrial biomedical platform.** Original Research Article. *Biochimica et Biophysica Acta (BBA) - Molecular Cell Research*, Available online 31 October 2015.

(Cont'd on page 14)

Awareness of Barth Syndrome Continues to Grow

(Cont'd from page 13)

2. Pokorná L, Cermáková P, Horváth A, Baile MG, Claypool SM, Griac P, Malínský J, Balážová M. **Specific degradation of phosphatidylglycerol is necessary for proper mitochondrial morphology and function.** *Biochim Biophys Acta.* 2015 Oct 19;1857(1):34-45. doi: 10.1016/j.bbabi.2015.10.004. [Epub ahead of print].
3. Mathur A, Ma Z, Loskill P, Jeeawoody S, Healy KE. **In vitro cardiac tissue models: Current status and future prospects.** *Adv Drug Deliv Rev.* 2015 Sep 30. pii: S0169-409X(15)00214-8. doi: 10.1016/j.addr.2015.09.011. [Epub ahead of print] Review.Δ
4. Gaspard GJ, McMaster CR. **Cardiolipin metabolism and its causal role in the etiology of the inherited cardiomyopathy Barth syndrome.** *Chem Phys Lipids.* 2015 Sep 25. pii: S0009-3084(15)30054-2. doi:10.1016/j.chemphyslip.2015.09.005. [Epub ahead of print] Review.*
5. Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** *Pediatr Cardiol.* 2015 Sep 4. [Epub ahead of print]Δ
6. Shen Z, Ye C, McCain K, Greenberg ML. **The role of cardiolipin in cardiovascular health.** *Biomed Res Int.* 2015;2015:891707. doi: 10.1155/2015/891707. Epub 2015 Aug 2. Review.*
7. Planas-Iglesias J, Dwarakanath H, Mohammadyani D, Yanamala N, Kagan VE, Klein-Seetharaman J. **Cardiolipin interactions with proteins.** *Biophys J.* 2015 Aug 20. pii: S0006-3495(15)00769-9. doi: 10.1016/j.bpj.2015.07.034. [Epub ahead of print]
8. Gifford SM, Meyer P. **Enzyme function is regulated by its localization.** *Comput Biol Chem.* 2015 Aug 8. pii: S1476-9271(15)30112-2. doi: 10.1016/j.compbiolchem.2015.08.004. [Epub ahead of print]
9. Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** *J Multidiscip Healthc.* 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review.
10. Chin MT, Chien WM, Dinca A. **Modified tafazzin proteins and methods of making and using the same.** Patent Application 20150203827. July 23, 2015. (Patent Application)
11. Tocchi A, Quarles EK, Basisty N, Gitari L, Rabinovitch PS. **Mitochondrial dysfunction in cardiac aging.** *Biochim Biophys Acta.* 2015 Jul 17. pii: S0005-2728(15)00152-8. doi: 10.1016/j.bbabi.2015.07.009. [Epub ahead of print]
12. Wilson D. **Methods and compositions for the prevention or treatment of Barth syndrome.** International Publication Number WO 2014/134554 A1. 2014 Sep 4. (Patent Application)
13. Stepanyants N, Macdonald PJ, Francy CA, Mears JA, Qi X, Ramachandran R. **Cardiolipin's propensity for phase transition and its reorganization by dynamin-related protein 1 form a basis for mitochondrial membrane fission.** *Mol Biol Cell.* 2015 Jul 8. pii: mbc.E15-06-0330. [Epub ahead of print]
14. Grevengoed TJ, Martin SA, Katunga L, Cooper DE, Anderson EJ, Murphy RC, Coleman RA. **Acyl-CoA synthetase 1 deficiency alters cardiolipin species and impairs mitochondrial function.** *J Lipid Res.* 2015 Aug;56(8):1572-82. doi:10.1194/jlr.M059717. Epub 2015 Jul 1.
15. Ikon N, Su B, Hsu FF, Forte TM, Ryan RO. **Exogenous cardiolipin localizes to mitochondria and prevents TAZ knockdown-induced apoptosis in myeloid progenitor cells.** *Biochem Biophys Res Commun.* 2015 Jul 8. pii: S0006-291X(15)30243-6. doi: 10.1016/j.bbrc.2015.07.012. [Epub ahead of print]*
16. Nagueh SF, Zoghbi WA. **Role of imaging in the evaluation of patients at risk for sudden cardiac death: Genotype-phenotype intersection.** Review Article. *JACC: Cardiovascular Imaging, Volume 8, Issue 7, July 2015, Pages 828-845.*
17. Sauerwald J, Jores T, Eisenberg-Bord M, Chuartzman SG, Schuldiner M, Rapaport D. **Genome-wide screens in yeast highlight a role for cardiolipin in biogenesis of mitochondrial outer membrane multispan proteins.** *Mol Cell Biol.* 2015 Jul 6. pii: MCB.00107-15. [Epub ahead of print]
18. Angelini R, Lobasso S, Gorgoglione R, Bowron A, Steward CG, Corcelli A. **Cardiolipin fingerprinting of leukocytes by MALDI-TOF/MS as screening tool for Barth syndrome.** *J Lipid Res.* 2015 Jul 5. pii:jlr.D059824. [Epub ahead of print]*Δ
19. Ye Y, Carlsson G, Karlsson-Sjöberg JM, Borregaard N, Modéer TU, Andersson ML, Pütsep KL. **The antimicrobial propeptide hCAP-18 plasma levels in neutropenia of various aetiologies: A prospective study.** *Sci Rep.* 2015 Jun 29;5:11685. doi: 10.1038/srep11685.
20. Morita SY, Terada T. **Enzymatic measurement of phosphatidylglycerol and cardiolipin in cultured cells and mitochondria.** *Sci Rep.* 2015 Jun 30;5:11737. doi: 10.1038/srep11737.
21. Cadalbert LC, Ghaffar FN, Stevenson D, Bryson S, Vaz FM, Gottlieb E, Strathdee D. **Mouse tafazzin is required for male germ cell meiosis and spermatogenesis.** *PLoS One.* 2015 Jun 26;10(6):e0131066. doi: 10.1371/journal.pone.0131066. eCollection 2015.*
22. Ryan RO. **Metabolic annotation of 2-ethylhydracrylic acid.** *Clin Chim Acta.* 2015 Jun 23. pii: S0009-8981(15)00295-8. doi: 10.1016/j.cca.2015.06.012. [Epub ahead of print] Review.*
23. Huang Y, Powers C, Madala SK, Greis KD, Haffey WD, Towbin JA, Purevjav E, Javadov S, Strauss AW, Khuchua Z. **Cardiac metabolic pathways affected in the mouse model of Barth syndrome.** *PLoS One.* 2015 Jun 1;10(6):e0128561. doi: 10.1371/journal.pone.0128561. eCollection 2015.
24. Hijikata A, Yura K, Ohara O, Go M. **Structural and functional analyses of Barth syndrome-causing mutations and alternative splicing in the tafazzin acyltransferase domain.** *Meta Gene.* 2015 Apr 22;4:92-106. doi: 10.1016/j.mgene.2015.04.001. eCollection 2015 Jun.
25. Melser S, Lavie J, Bénard G. **Mitochondrial degradation and energy metabolism.** *Biochim Biophys Acta.* 2015 May 12. pii: S0167-4889(15)00156-1. doi: 10.1016/j.bbamcr.2015.05.010. [Epub ahead of print] Review.
26. Taylor D, Brady JE, Li G, Sonty N, Saroyan JM. **Characterization of pain in patients with Barth syndrome.** *Children's Health Care, DOI: 1080/02739615.2014.996882.Δ*

Power of Kindness

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Nagel, Robin
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Neier, Melissa
Neira, Beatriz
Nelson, Lisa
Nesbitt, Carol
Nicoll, Rev. Tom & Mary
Northrup, Bruce & Jan
O'Connell, Ryan & Janet
Odouard, Francois & Reshmi
O'Hara, Peter & Sabina
O'Keefe, Linda
Olson, Dick & Sharon
O'Shaughnessy, Jim & Missy
Ouellette, Monique
Palmer, Kirk & Lori
Pannell, Katherine
Panno, Dr. David & Leelee
Paolini, Ann
Pardo, Marilyn
Parter, Kathryn

(Cont'd on page b)

Power of Kindness

(Donor categories are based upon the past 18 months of cumulative giving from 5/1/2014–12/2/2015)

(Cont'd from page a)

Pattee, Diane
 Perry, Najjiyya
 Peterson, Carl & Andee
 Phillips, Nancy & Tom Dunne
 Phoon, Dr. Colin & Janet
 Pierson, Frank & Nancy
 Pierson, Dr. Richard & Kitty
 Plotkin, David
 Plumez, Jean Paul & Dr. Jackie
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 Purcell, Bob & Jackie
 Purdy, Dr. Bill & Molly
 Rabkin, David
 Rader, Stephanie
 Ratner, Ian & Carol
 Raynaud, Veronique
 Reid, Colin
 Rey, Lisa
 Riecker, Kristine
 Rigney, Joseph
 Roberts, John & Betty
 Robidoux, Monique
 Rodamer, Bob
 Rodbell, Arthur & Rhoda
 Rodbell, Gary & Colette
 Rodbell, Julia
 Rodbell, Michael & Margaret
 Rodbell, Mitchell & Liz
 Rogers, Larry & Catherine
 Roggow, Beth
 Rosenblatt, Stephen
 Rosenfeld, Lisa
 Rosenshine, Jon & Mary Pat
 Rotondi, Andrew & Mary Frances
 Rozansky, Marc & Sherri
 Ruderman, Gail
 Ruderman-Fennelly, Nancy
 Russell, Harold & Margo
 Russell, Paul & Sara
 Rutherford, Alanna
 Rutledge, Elizabeth
 Sadler, Evelyn
 Sakakeeny, Brandt
 Sanborn, Peter
 Sanford, Katherine
 Santamaria, Marco & Judy
 Schlapak, Sonja & Gregor
 Schreiber, Mary Ann & Ed Mooney
 Schwendener, Paul & Barbara Gessler
 Seay, Laura
 Segal, Mark & Anne
 Segal, Mark & Heather
 Sherer, Peter & Marilu
 Sherer, Tony
 Shtull, Ora
 Sims, Mac & Joanne
 Singer, Craig
 Singer, Scott & Mary Jo Romano
 Sleeper, Emmy Lou
 Smith, Mike & Susan
 Smyser, Georgiana
 Snedeker, Rob & Fran
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 Spaulding, Cici
 Staub, Amy
 Steinberg, Jack
 Strain, Fraser & Donna
 Strohl, Linda
 Sullivan, Barb
 Suzuki, Matthew
 Swain, Bob & Virginia
 Swennen, Erik & Veerle
 Symington, Jane
 Tecklin, David
 Thach, Christy
 The Rye Tri Club Inc.
 Thomas, Stephen
 Tillinghast, Pamela
 Townley, Birgit
 Tsai, Lynn

Tunguz, Stefan & Julie
 Tutwiler, Temple & Lucy
 Vail, Clayton
 Van Valen, Colleen
 Van Valen, Ed & Jeanne
 Van Valen, Kera
 Varner, Judy
 Viebranz, Elaine
 Vourlekis, Melina
 Wallerstein, Diana
 Waterman, Chris & Marcy Wilkov
 Waters, Marty & Helen
 Watt, Richard & Gill
 Waugh, Emily
 Wayne, Sharon
 Weissman, Alan
 Werner-O'Brien, Claire
 Weston, Garrett
 White, Bill & Marian
 Wilkins, Sue & Dr. Mike
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 Wofford, Daniel
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 Zierk, Holly
 Zierk, Tom & Gail

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 Buly, Dr. Bob & Lynne
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 Isaac, Paul & Karen
 Lummis, Marilyn
 Lummis, William & Dossy
 McCurdy, Steve & Kate
 Miller, Kirt & Martha
 Millet Encalada, Mario & Cecilia
 Pierson, Dr. Robin & Allene
 Russell, Dr. Paul
 Sanchez, Anthony
 Sernel, Gordon & Sharon
 Sernel, Marc & Tracy
 Shapiro, Brenda
 Smith, Leslie
 The Lebensfeld Foundation

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 American Express PAC Match
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 Barad, Seth & Amy
 Barth Syndrome Trust
 Barth Syndrome South Africa
 Bauer Family Foundation
 Bellig, Mike & Sara
 Bercovich, Bruce
 Blouir, Matthew & Anna
 Branagh, Thomas & Diane
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 Kirkbride, Brandon & Kayla
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 Lummis, Ghent & Ginger
 Lynn, Beth
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 McCurdy, Chris & Kris
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 Neill, Erik
 Olson, Ken & Tina
 Osnos, Suze & Peter
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 Randolph, Dr. Peter & Helen
 Shaw, Walt
 Singer, Steven
 Smith, Renee
 Stone, Denise
 Swennen, Erik & Veerle
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 Vaisman, Natan & Beth Roberts
 Van Langendonck, Karel & Magda
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 Zeller, Bill & Anne

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(\$50 - \$999)

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 Ameriprise Financial Employee Giving Campaign
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 Adams, Jamie
 Addington, Devin
 Alden, Alexis
 Alimanestianu, Alexander
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 Allman, Tom & Jane
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 Ammann, Sandra
 Anderle, Eric
 Anderson, Jeff
 Anderson, Tommy & Allana
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 Asaff, Ernie & Colette
 Arovas, Gregory
 Aviles, Michael
 Axelrod, Dr. David & Jessica
 Baer, Ben & Susie
 Baffa, Mary
 Baffa, Ted & Rosemary
 Baker, Timothy
 Ballard, Stacey & Frank
 Barab, Stuart & Anne
 Barnett, Linda
 Baron, John & Christine
 Barth, Dr. Peter
 Basler, Dr. Rod & Debbie
 Bater, Jennifer
 Bates, David
 Bauer, Steve
 Beckett, Robert
 Behnke, Eugene & Margaret
 Belfatto, Eileen
 Bell, Jenny
 Bellig, John & Susan

Bennett, Claudia
 Bennett, Judith
 Berens, Wayne & Harriet
 Berland, Leslie
 Bernal, Francisco
 Berry, Jim
 Berry, Rick & Carla
 Berry, Shane
 Bertan, David
 Bertling, Norbert & Donna
 Bettenhausen, Bob & Joanne
 Betz, Christopher & Megan
 Bierbaum, Frederick & Dionna
 Bilik, Kari
 Bingham, Dr. Dave & Kathy
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 Bittel, Rhonda
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 Bodary, Michael & Lisa
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 Bogert, Carroll
 Bohnert, Kaye
 Bosak, John & Anita
 Bosak, Mary
 Botto, Patrice
 Bougger, Annie
 Bowen, Michael
 Bowen, Shelley
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 Bowron, Ann
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 Branagh, Geoffrey
 Branagh, Jack & Lyn
 Branagh, Katie
 Branagh, Kimberly
 Brandt, Douglas
 Brause, Cheryl
 Brasseur, Tanya
 Brewster, Brinks & Laira
 Britt, Peggy
 Britton, Lori & Jimmy
 Brody, David & Jeanne
 Brody, Meghan
 Brooks, Cameron & Tawnya
 Bryant, Bobby & Pamela
 Bryant, Dr. Randall
 Buddemeyer, David
 Buddemeyer, Donna
 Buddemeyer, Leslie
 Bull, Dave & Sarah
 Burdick, Lindsey
 Burgan, Francine
 Burgher, Bedford
 Burke, Margaret
 Burmeister, Blaine & Abby
 Burmeister, Chuck & Marita
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 Burr, Richard
 Butera, Jacquie & Donald Kyle
 Bruno, Ellen
 Callahan, Lynn
 Calvert, Elaine
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 Campbell, Tonja
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 Canova Jr., Frank
 Capasso, Ruth Ann
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 Carroll Masterson, Marie
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 Cent, Beverly & Norbert
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 Cheatham, Joyce
 Cheatham, Linda
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 Cherniak, Miriam
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 Clark, Rebecca
 Cleek, Robin
 Cohune, Vicky
 Coleman, Charlie
 Collazzi, V.C.
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 Condon, Mark
 Congdon, Margot
 Consos, Joey
 Conway, April
 Coombe, Amy & Burt
 Cotters, George & Shelley
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 Couch, Rick
 Courtney, Shirley
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 Cracchiolo, James & Marilyn
 Crandall, Risa
 Crawford, Michael & Eleanor
 Cripps, Tia
 Cronin, Kathleen
 Crowley, Chuck
 Crowley, Diane
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 Crozier, Nancy
 Cuccia, Helen
 Cushmore, Patricia
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 Dastous, Brock
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 Davis, Elizabeth
 Davis, Elzak
 De Feis, Mary
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 Derusha-Mackey, Sarah
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 Drake, Laura
 Drake, Marcia
 Draper, Steve
 Drattell, Maria
 Drury, Paul
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 Dugan, Patricia
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 Dunn, Robert & Helen
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 Durando, Courtney
 Edgemon, Matthew
 Egueur, Madeleine
 Elliston, Ann
 Elmer, Madeleine
 Elwood, Lynn & Rick
 Engberg, Renee
 Enterline, Carol
 Epand, Dr. Richard & Mrs.
 Epstein, Paul & Leah
 Erdhaus, John & Sharron
 Evans, Robert & Beverly
 ExecComm LLC
 Fairchild, Chris

(Cont'd on page c)

Power of Kindness

(Donor categories are based upon the past 18 months of cumulative giving from 5/1/2014-12/2/2015)

(Cont'd from page b)

Fairchild, Dewayne & Julie
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 Ganz, Doug & Pam
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 Garcia, Michele
 Garcia, Rudy & Lin
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 Green, Frances
 Green, Mitch & Susan Yamaguchi
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 Gress, Emil & Natalie
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 Groff, Lindsay
 Grubb, Johnny & Jane
 Grzesiak, Lisa
 Gungor, Umut
 Gunther, Nicole
 Hackett, Christopher & Gina

Haessler, John & Nancy
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 Hall, James & Amie
 Hallmark Agency, Inc.
 Hallam, Ken & Theresa
 Hansen, Stephanie
 Happy Tails
 Harkness, Tyler
 Harman, James & Merlene
 Harris, Mark
 Haviland, Jordan
 Hedgecock, Norm & Debbie
 Heilmann, Kathy
 Heller, Cindy
 Helms, Kathryn
 Hendrick, Susan
 Hendricks, Dr. Joe
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 Holly, Kristi
 Holly, Peggy
 Holly, Ron
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 Horluc, Todd & Jennifer
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 Jarecha, Paul
 Jarrett, Cate
 Jefferies, John & Shari
 Jeffery-Miller, Dorothy
 Jensen, Tom & Marilyn
 Joens, Steve
 Jofre, Jaime & Laura
 Johnson, Jennifer
 Jones, Kristy
 Kahn, Natasha
 Kalapasev, Ned & Brie
 Kaminer, Riley
 Kaminsky, Gila
 Karp, Joseph
 Kaufman, Jeff & Wendy
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 Kelly, Seth
 Kennedy, Dianne
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 Lascurettes, Nancy
 Lascurettes-Mangiapane, Vince
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 Lauer, Valerie
 Law, Oliver & Susan
 Lawson, Richard
 Le Jamtel, Jerome
 Lederman, Dr. Seth & Sarah
 Lee, Richard
 Lending Club
 Leser, Robert & Hilary
 Lever, Charles & Beverly
 Levinson, Charles
 Levinson, Marla
 Liebenguth, Mouse
 Lieberman, Amy
 Lind, Michael
 Linder, Dr. Max & Pat
 Lindquist, Douglas & Patti
 Loftus, P.
 Logan, Christie
 Lollar, Mr. Lollar
 Long, David
 Lord, Sarah
 Lynch, Lilian
 Lyon, Janie
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 MacKay, Douglas
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 Mahoney, Sara
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 Marsico, Michael
 Martin, John
 Martin, Melanie
 Mask, Harold & Gayle
 Massengale, Dr. Martin & Ruth
 Massie, Kaylan
 Matelich, George & Susan
 Maynard, Dr. Ed & Lisa
 Maynard, Marilyn
 McAnally, Zac
 McClellan, MGC, Rebecca
 McCormack, Helen
 McCormack, Timothy & Marcia
 McCracken, Paul
 McFee, Wendy & Jmel Wilson
 McHugh, Laura
 McKay, Richard
 McNair, Gail
 McNally, Mark
 McNeil, Thomas & Michelle
 McPhee, Kristina
 McTear, John & Donna
 Mellen, Ann
 Melrose, David & Pat
 Menkhaus, David
 Merrill, Allan
 Michaud, Steven & Yoko
 Michener, Sandt & Kathryn
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 Mihalich, Jane
 Miller, Denise-Aguilar
 Miller, Jane & Clyde Rayner
 Miller, Jill
 Millman, Paul & Susie

Miloff, Michael
 Minnick, Gates & Daisy
 Montanaro, Louis & Theresa
 Montgomery, John
 Moore, Nigel & Lorna
 Moreland, Dr. Brian & Rachel
 Morey, Dale & Bonnie
 Morgan, Anne
 Morgan, Derek & Laura
 Morgan, Kathleen
 Morock, Kim
 Morris, Erin
 Moskowitz, Rebecca
 Mouracade, Valerie
 Muaddi, Susan
 Murphy, Donald & Jane
 Myers, Edward & Anita
 Nachman, David
 Neece, George & Sue
 Neece, Kate
 Neece, Michael
 Neece, Rebecca
 Neece, Sarah
 Neece, Sue
 Neece, Thomas
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 Neill, Merrily
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 Nelson, Scott & Teri
 Nelson, Wayne & Joyce
 Nemanic, Erik & Kathy
 Nevel, Bonnie
 Newacheck, Rick & Pamel
 Nicoll, Rev. Tom & Mary
 Northrup, Bruce & Jan
 Nowak, Tanya
 Nurkowski, John
 Oates, Jason
 Obalde, Manuel & Carla
 O'Connell, Ryan & Janet
 O'Connor, Mary
 Odell, Heather
 Odoardi, Anne
 Odouard, Francois & Reshmi
 O'Keefe, Ann
 Olsen, Steven & Nancy
 Olson, Amy
 Olson, Donald & Eunice
 Olson, Loy & Julie
 Olson, Dick & Sharon
 O'Rourke, Courtney
 Orso, John & Theresa
 Osterhaus, Greta
 O'Sullivan, John
 Pacak, Dr. Christina
 Pagano, James & Mary Lou
 Palmer, Kirk & Lori
 Pannell, Katherine
 Paolizzi, Vincent & Celeste
 Papone, Aldo & Sandra
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 Parlato, Carolyn
 Peck, Glenn
 Peck, Susan
 Pellettieri, Ginger
 Pena, Chris & Kristi
 Pepworth, Donald
 Percival, Cathy & Steve
 Pergam, Carl & Jeannette
 Perusco, Ashley
 Pfizer Foundation Matching
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 Phillips, Bill
 Phillips, Mark & Katherine
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 Poe, Kim
 Powell, Brian
 Prendergast, Sheila
 Presler-Marshall, Martin
 & Elizabeth
 Puhr, Edward & Kathryn
 Purcell, Bob & Jackie

Radelat, Troy
 Rader, Carol
 Rader, Stephanie
 Rainier, Nancy
 Randell, Dr. Jay & Amer
 Rath, Gene
 Ratner, Ian & Carol
 Rawley, Ann
 Ren, Dr. Mindong
 Renbarger, Sam
 Rhodes, Alberta
 Rigsby, Robert & Deborah
 Rimby, David
 Ritter, Chris & Cathy
 Rizedesign
 Roberts, John & Betty
 Robinson, Kyle
 Robinson McFarland, Jane
 Rodamer, Bob
 Rodbell, Arthur & Rhoda
 Rodbell, Gary & Colette
 Rodbell, Michael & Margaret
 Rodbell, Mitchell & Liz
 Rollins, Michael & Kim
 Rorschach, Bob & Susan
 Rosenblatt, Stephen
 Rosenfeld, Lisa
 Roskens, Dr. Ronald & Lois
 Rosson, Andrew
 Roth, David
 Rothschild, Adam & Kathy
 Rowlett, Michael
 Rozbruch, Robert & Yonina
 Russell, Gary
 Russell, Harold & Margo
 Russell, Paul & Sara
 Ryan, Patricia
 Ryan, Robert
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 Saporito, Gennaro & Alisa
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 Schmitt, Robert
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 Schroeder, Wallace
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 Segal, Mark & Heather
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 Sellet, Rebecca & David
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 Simon, Thomas & Nancy
 Slawson, Kirsten
 Smith, Dr. Herbert
 Smith, Jeff & Judi
 Smith, Jim & Mary Jo
 Smith, Richard & Toni
 Smith, Tonia
 Solomon, Lauren
 Sonderegger, Ted & Mary Ann
 Sorrento, Nora
 Spalla, Katie
 Sparks, Kara
 Spencer, Louise
 Spotts, Dr. Jules

(Cont'd on page d)

Power of Kindness

(Donor categories are based upon the past 18 months of cumulative giving from 5/1/2014–12/2/2015)

(Cont'd from page c)

St. Joseph Notre Dame High School	Sweetman, Tina	Unger, Evelyn	Wells, Anna	Wood, Kelli
Staub, Amy	Taddeucci, Katie	United Way California Capital Region	Weltlich, Bob & Dodie	Woodward, Gordon & Ann
Staudt, Dennis & Patty	Taegtmeier, Heinrich		Weltlich, Mark	Woodward, Rick & Lina
Stenson, Marie	Takata, Gregg	Urmey, William & Tamara	Weltlich, Steve & Sharon	Woodward, Steven
Stenson, Michael	Taylor, Paul	Vail, William	Wendling Orthodontics	Wright, Cameron & Jessica
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Sterling, James	Tegt, Dr. Tom & Barb	VanRensselaer, Cheryl	Wertlieb, Linda	Wynn, Ken
Stevens, Sandra	Temby, Michelle	Varner, Tom & Beth	West, Shilo	Yon, Kirk
Stevenson, Robert & Sharon	Teodecki, Amanda	Vernon, Aaron	Wharton, Philip & Philippa	Young, Penny
Stewart, Lori	Thach, Christy	Viegas, Martin	Wiederspan, Mark & Jess	Yu, Huayun
Stohs, Dr. Gene & Kristen	Thames, Sidney	Vogt, Jerre	Wilkins, Dr. Jerry	Zajac, Adina
Stone, Jeffrey & Vicky	Thompson, Art & Carol	Wadsworth, Kimu	Wilkins, John	Zeitner, Eric
Stone, Kimberly	Thompson, Barbara	Wagner, Doreen	Wilkins, Dr. Lee & Kristy	Zoland, Yale & Wendy
Stoner, David & Teresa	Thompson, Nathan	Walden, Leslie	Wilkins Burnham, Joanne	
Strain, Fraser & Donna	Thomson Reuters Matching Gifts	Waller, Scott	Williams, Anne	
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Stuckey, Dennis & Nancy	Tizzano, Alissa	Walton, Darren	Williams, Jennifer	
Stuhreyer, Tom & Kim	Torbert, Tracy & Jay	Waters, Marty & Helen	Williams, Kathleen & Suzi Anderson	
Stuhreyer, William & June	Toth, Dr. Matt & Marilyn	Watkins, Becky & David George	Wind, Kathleen	
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Sullivan, Tim	Townley, Birgit	Wayne, Sharon	Winters, Ian	
Sutherland, Laura	Trimmer, Lori	Weigel, Larry & Patricia	Withem, Angela	
Swabe, Carolyn	Tunguz, Stefan & Julie	Weinstock, Sharon	Wolff, Patricia	
Swabe, Rod	Turull, Albina	Weissman, Alan		

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Atwell, Joshua & Morgan	Dane, BS, Brittany	Kagan, Dr. Valerian E.	Neece, Michael	Theda, Dr. Christiane
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Braverman, Dr. Nancy	Galbraith, Lois	Lorberboum-Galski, Prof. Haya	Ren, Dr. Mindong	Wilkins, John
Broadridge	Ganote, Felicia C.	Lou, Wenjia	Reynolds, Dr. Stacey	Wilkins, Sue
Brown, Dr. David A.	Garratt, DClinPsych, Vanessa	Lu, Ya-Wen	Riddiford, CNS, Debbie	Woodward, Kevin
Bruno, Ellen	Garry, Dr. Daniel	Lumms, Ghent	Riss, Shoshanna	Woodward, Stacey
Bryant, Dr. Randall	Gawrisch, Dr. Klaus	Mancino, Michael	Ritter, RN, Catharine L.	Ye, Cunqi
Buddemeyer, Randy	Gillum, Dr. Matthew P.	Mann, Sheila	Rodbell, Gary	Yin, Dr. Huiyong
Buly, Dr. Bob & Lynne	Goncalves, Dr. Renata	Mann, Dr. William	Rodbell, Julia	Yu, ScD, Xin
Buly, Jocey	Gonzalez, Dr. Iris L.	Mannes, Florence	Rossano, Dr. Joseph W.	Zaragoza, Dr. Michael V.
Burgess, OTS, M. Emily	Gorecki, Michelle	Mannes, Philippe	Ryan, Dr. Robert O.	Zeller, Anne
Byrne, Dr. Barry J.	Greenberg, Dr. Miriam	Manton, Annick	SMART Group (NHLBI – NIH)	Zeller, Bill
Cade, Dr. W. Todd	Grevengeod, Trisha J.	Marshall, Dr. Bess	Schlame, Dr. Michael	Zhang, Dr. Ji
Cazzaniga, Dr. Paola	Groff, Lindsay	McClellan, MGC, Rebecca L.	Sedefian, Lynda	
Chicco, Dr. Adam	Groft, Dr. Stephen C.	McCormack, Susan	Segal, Heather	
Chin, Dr. Michael T.	Hagar, Elissa	McCurdy, Eliza	Sernel, Marc	
Clayton, Nicol	Hatch, Dr. Grant M.	McCurdy, Kate	Shapiro, Heller An	
Claypool, Dr. Steven	Higgins, Jack	McCurdy, Steve	Shu-Zhe, Dr. Ding	
Cole, Dr. Laura	Hone, Susan	McKown, Chris	Smoot, Dr. Leslie B.	
Collier, BA, Amanda	Hope, Chris	Mejia, Edgard M.	Stevens, Sandra	
Congenital Cardiology Today	Hope, Michael	Metallo, Dr. Christian	Steward, Prof. Colin G.	
Conway, Dr. Laura J.	Houten, Dr. Sander	Millay, Dr. Douglas	Stewart, Dr. Douglas R.	
Coombes, Prof. Jeff	Huang, MS, Yu-Yun	Mitchell, Dr. Sabrina	Strathdee, Prof. Douglas	
Corcelli, Dr. Angela	Huffman, Dr. Kim M.	Moore, Lorna	Strauss, Dr. Arnold W.	
	Huizing, Dr. Marjan	Moore, Nigel	Stutts, Dr. Lauren A.	

Barth Syndrome Researchers Awarded RO1 Grants from the National Institutes of Health



Dr. William Pu

William T. Pu, MD, Boston Children's Hospital, Boston, MA, has been awarded an RO1 grant from the National Institutes of Health (NIH) entitled "**Understanding mitochondrial regulation of cardiac development and function through studies of Barth syndrome.**" (Project #: 1R01HL128694-01)

Abstract Text:

Heart muscle cells contain the highest concentration of mitochondria in the body. While it is well recognized that these mitochondria are required to produce ATP needed for the adult heart to sustain its pumping activity, other roles of mitochondria in the development and function of the heart are less well understood. Mitochondrial disorders, such as Barth syndrome (BTSH), cause left ventricular non-compaction, a form of heart disease in which the wall of the heart is not properly formed. They also can cause heart muscle dysfunction without causing energy depletion. By studying the disease processes involved in BTSH, this study will illuminate the function of mitochondria in heart development and function. The study will use our understanding of Barth syndrome pathogenesis to perform pre-clinical testing of new treatment strategies for this disease.



Dr. Michael Schlame

Michael Schlame, MD, New York University School of Medicine, New York, NY, has been awarded an RO1 grant from the National Institutes of Health (NIH) entitled "**Abberant cardiolipin dynamics in Barth syndrome.**" (Project #: 1R01GM115593-01)

Abstract Text:

Barth syndrome (BTSH) is a disorder of the mitochondrial metabolism of lipids, in particular the mitochondria-specific lipid cardiolipin, and thus provides unique opportunity to address this gap in a context relevant to human health. The objective of this application is to identify the mechanism that causes partial replacement of cardiolipin by monolyso-cardiolipin in BTSH and to elucidate its functional consequences. This objective fits into our broad goals to understand the function of cardiolipin in mitochondria and to unravel the molecular pathophysiology of BTSH. The proposed project will provide critical insight into the role of mitochondria in cardiac differentiation and demonstrate the effect of two drugs in a mouse model of the disease. The proposed study is significant because it will establish the molecular pathogenesis of BTSH and it will test a potential therapy of the disease in a mouse model.

Opportunity to Participate in Research

Dr. John Lynn Jefferies of Cincinnati Children's Research Foundation is doing a research study concerning the assessment of quality of life, anxiety, and depression in Barth syndrome. Please consider the information below and contact Dr. Jefferies directly if you decide to help.

Do You or a Loved One Have Barth Syndrome?

CCHMC IRB # 2014-7162: V1

What: We want to better understand the frequency of depression, anxiety and health related quality of life in people with Barth syndrome (BTSH). We also want to develop a patient/parent-reported, BTSH-specific checklist that will help us identify gaps in clinical care.

Why: We hope this information can be used to learn more about targets for clinical interventions and treatments.

Who: Children, teens and adults, five years and older, who have Barth syndrome may be eligible for participation. Participants must have home internet connection to complete online questionnaires.

What's Involved: Parents/participants will complete three questionnaires, six months apart, as well as take part in a focus group with other participants and clinical experts to develop and test a BTSH-specific symptom checklist.

Pay: Participants will not be paid for their time, travel or effort during this research study.

Contact: 513-803-0366 or hirc@cchmc.org

SAVE THE DATE!

8th International Scientific, Medical & Family Conference Team Barth

July 18-23, 2016

Hilton Clearwater Beach Resort, Clearwater, Florida, USA

Save the date! The 2016 Barth Syndrome International Scientific, Medical & Family Conference is scheduled for July 18-23, 2016 at the Hilton Clearwater Beach Resort located in Clearwater, Florida. The hotel is right on the beach with two pools, many restaurants, ample shopping, and exciting activities all within walking distance.

Call for Speaker Abstracts

The Barth Syndrome Foundation (BSF) is soliciting speaker abstracts for the Scientific & Medical Sessions of the 8th International Barth Syndrome Scientific, Medical & Family Conference. The Sci/Med sessions will take place on Thursday and Friday, July 21st and 22nd, and presentations should cover clinical and scientific areas directly related to Barth syndrome. Invited speakers will have 30 minutes to present. **The deadline for Speaker Abstract submission is March 1, 2016.** Please submit your Poster Abstract to Matthew Toth at: Mtothbsf@comcast.net or matthew.toth@barthsyndrome.org.

We expect to invite ~ 18 speakers to describe their work caring for Barth syndrome individuals, or how their work furthers the goal of finding a specific treatment for Barth syndrome or addresses aspects that relate to the pathophysiology of Barth syndrome. In order to have a fair and orderly process, we are asking the potential speakers to provide us with a one-page abstract (less than 400 words) describing in general terms what they will present. Based on these abstract submissions we will choose the speakers and assemble the agenda for the SciMed sessions. The Barth Syndrome Foundation will provide travel expenses, hotel accommodations, and common meals for the invited speakers, unless they are able to generously use their own funds for this purpose. In addition, the 2016 Conference will have a Poster session (on Thursday afternoon) along with a Poster stipend program that can help defray the cost of attendance if one is not invited as a speaker. For 2016 we expect to ask four poster presenters to speak at the Friday afternoon session for 15 minutes each.

Call for Poster Abstracts

The Barth Syndrome Foundation 2016 Scientific and Medical Conference Organizing Committee (COC), comprised of members of the Barth Syndrome Foundation international Scientific & Medical Advisory Board, invites the submission of abstracts for poster presentations related to the scientific and/or clinical aspects of Barth syndrome. **The deadline for Poster Abstract Submission is May 1, 2016.** All submitted abstracts will be peer-reviewed by the COC. Once accepted, the submitting author will be expected to present his/her corresponding poster at a specific time during the Conference. Please submit your Poster Abstract to Matthew Toth at: Mtothbsf@comcast.net or matthew.toth@barthsyndrome.org.

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

Scholarship Program

The Barth Syndrome Foundation offers a limited number of travel scholarships for qualifying physicians, clinical residents/fellows/students, nurses, and other allied health professionals to help defray the cost of attending the 2016 Conference. This program is designed to encourage medical practitioners to increase their knowledge about and improve their care of Barth syndrome individuals. Program and application information will be available at www.barthsyndrome.org.

Why You Need To Attend:

"My attendance at BSF's conference was invaluable in learning about patients with this disorder and about scientific progress into the mechanisms of disease and genotype-phenotype correlations." ~ Arnold W. Strauss, MD, BK Rachford Professor and Chair, Department of Pediatrics, University of Cincinnati College of Medicine; Director, Cincinnati Children's Research Foundation; Chief Medical Officer, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio

Barth Syndrome Foundation Affiliates



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Board of Directors

Paola Cazzaniga, President
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Margherita Usai

Inside Barth Syndrome Trust's Fundraising

By Nigel Moore, Trustee, Barth Syndrome Trust

I am always amazed by the commitment, efforts and endurance of our families, fundraisers, volunteers and donors. The United Kingdom is far removed from charity heaven these days, and our fundraisers face difficulties with good humour and perseverance.

Fundraising is dominated by big charities which have substantial public recognition. It also sometimes gets a bad press because of some malpractice, bankruptcy, misuse of funds and aggressive techniques. The public perception is, therefore, not always favourable and we can all be "tarred with the same brush."

Times are tough, limiting the money available for donations. There are also so many good causes that it is difficult for people to know how to spread their giving. So the task of fundraising by and for a small, little known charity with limited resources is much harder. On the upside though, our fundraisers are all linked by their affection and admiration for someone who has or is battling Barth syndrome, and this makes it personal and heartfelt.

We strive to make a difference for our families and for those whose lives are affected by Barth syndrome. Each donation, each fundraiser, no matter how small, brings us all nearer to our goals. It gives me great pleasure to record the work of some of our volunteers and donors. Thank you to all for your kindness. I'm sorry space does not permit mentioning everyone, but if you have done a fundraising event recently for us and your name is not mentioned below, please contact us as we would be delighted to include you in the next edition. We are always on the lookout for some more inspirational stories.

Donations

We are grateful for donations from families and friends in UK, Belgium and Ireland and have been nominated by staff at companies in various parts of the UK as a chosen charity. More donors have signed up for standing orders, and it is really helpful to have this regular income. We might not mention you by name here, but we know who you are and we depend on your often substantial personal donations. Thank you.

Eclipse Proclaim Personal Injury Awards

Thank you so much to Eclipse Legal Systems, who for the third time nominated the Barth Syndrome Trust (BST) as the charity of the evening at one of their award ceremonies. The Eclipse Proclaim Personal Injury Awards 2015 was held on 26 November at the Grange, St. Paul's Hotel in London. The awards ceremony was organised by Barker Brooks and headline sponsored by Eclipse Proclaim. Comedian Stephen K. Amos hosted the event. We thank Eclipse and their four hundred guests for their support.

Ever since they first heard about Barth syndrome through one of their employees — Alan Brown, whose four-year-old son Philip died of the condition in 2009 — staff at Eclipse Legal Systems have raised thousands of pounds for BST through sponsored events and at the awards ceremonies. We would like to thank them and their guests for their very generous donation of £1439, raised at the Eclipse Proclaim Personal Injury Awards, in memory of Philip Brown.



Dolores Evelyn of Eclipse Legal Systems & Comedian Stephen K. Amos with a cheque for BST
(Photo courtesy of BST 2015)

Hitting the Water and Road (The Vitruvian Triathlon) for Barth, by Matt Riley

At the start of 2015 I decided to take on one of the toughest physical challenges I have faced, a half Ironman triathlon. I entered the Vitruvian event at Rutland Water at the end of August, giving me eight months to get in shape for a 1.2 mile open water swim, 52 mile bike ride and a 13.1 mile run. During my training I decided to raise some money for the Barth Syndrome Trust in memory of a friend's child, Sebastian, who passed away in December 2013 from the disease, just 2 years old.

After eight months of solid training and a few warm up events, the day arrived and my alarm went off at the ridiculous hour of 2:30 am. Travel from home to Rutland Water followed and then setting up my station for the day, checking and double checking my packing list. As the sun rose I entered the water at 6:20 am for the swim with about 100 other competitors in my wave. There was a bit of a tussle for position, but I



Matt Riley beats his target
(Photo courtesy of BST 2015)

(Cont'd on page 19)

Inside Barth Syndrome Trust's Fundraising

(Cont'd from page 18)

managed to find a bit of space and got around the two laps quicker than I expected. Out of the water and then on to the bike, two laps around Rutland Water, a fairly hilly route and starting to get warm now.

Off the bike and on to my favourite element, the run. As a keen runner, I was happy to have this discipline last as I managed to make up quite a few places. I crossed the line after 5 hours and 37 minutes of constant effort, a massive relief and amazed that I beat my 5 hours 45 minute target. One of the toughest things I have done, but worth it to raise some money for a fantastic cause, having raised over £425.

Surprise Donations

STAGES, Shorne Theatre Appreciation & Group Entertainment Society, sent a very kind letter of appreciation for our work and a cheque for £250. Thank you to members for accepting the nomination of the Barth Syndrome Trust for this donation from surplus funds. STAGES organise social outings to theatres etc. for those living in the Shorne, Higham and Chalk areas in Kent.

Thank you to staff at Davies Turner for nominating BST for a third time to receive the donations of £187 from a recent Dressdown Friday.

Suzy Green's Fundraising

Suzy Green, friends and neighbours have again been busy. Here are some examples of their generosity:

- A Cheese and Wine party at friend Tina's home with a houseful of village mums was a great success, raising £95
- Jane Petrucci ran the North Norfolk Coastal Marathon over the wet and windy shingle, raising £435.00 so far (www.justgiving.com/Jane-Petrucci)
- £100.00 came from a raffle among friends at the home of neighbour, Sue
- Suzy's Facebook sales of donated goods amounted to £90
- A "bucket shake" at the Fordham Car Boot sale collected £601

Thank you for donations from a collection box at In a Spin, Fordham Pre-school and Whittlesford Social Club.

Fine Time at Bat and Ball Club

By Terri Allison, Volunteer, Barth Syndrome Trust

The Bat and Ball Club is a group of table tennis players from all over the UK who meet each year in November in Guernsey and then Jersey to play in two tournaments on each island. Following what was not our most successful table tennis tour of Guernsey and Jersey, in terms of winning, our final evening's entertainment of the much awaited "fines" for all players and supporters was a resounding success and very beneficial for BST.

This wonderful group of friends allowed themselves to be "prosecuted" collectively to the sum of £325. When you consider that fewer than 30 people contributed, it bears testament to their generosity and support of "our boys." Reasons for fines ranged from "wearing a flat cap outside of the North" to "being last down to breakfast." The usual heated denials were bandied about in defence but no one listened, and it only added to the fun of what is now the means of heartfelt annual donations from very caring benefactors. Touchingly, one player who was unable to join us sent his fines donation via a team mate. As always, I am delighted, grateful and privileged to be part of such a club.

I'd like to give a special mention to Sarah Whithorn who is the collator of these dubious misdemeanours and also runs the evening by reading out the "charges" to each individual. Previously, the fines were only levied on the players and a few pounds of small change would be donated to whatever charity box was on the bar in the hotel. Now, however, everyone who crosses Sarah's path, whom she considers will pay up, gets a penalty. Players, supporters and even some very good natured opposition friends contribute.

Thank you all never seems enough but thank you very, very much again.

Highly Professional Care Saved My Son

By Veera, Mother of Alex (age 5 months), United Kingdom

We are a Russian family from Estonia. My name is Veera and my husband's name is Gleb. This year our family grew when our son, Alex, was born on 19th June 2015. He was diagnosed with Barth syndrome five weeks afterwards.

I had chosen to have an elective Caesarean section for delivery. It was only after the diagnosis I realised how lucky we were to find a consultant who agreed to take responsibility, without medical concerns, for performing the major operation. I have a family history strongly suggestive of Barth syndrome. I didn't know about this at the time, and the results of my tests are still not available. My mother lost three sons and her sister lost two sons in the seventies in rural Russia. All my family is generally in good health, and I couldn't imagine that these stories could have an impact on me.

Alex was admitted to the neonatal unit with extremely low blood sugar (0.7 mmol/L) three hours after delivery, and he was stabilised only after three days. At the same time, my local doctors contacted the Metabolic team at Evelina Hospital (London) and started running some genetic tests. However, Alex's lactate was intermittently high, and we were transferred to Evelina Hospital a week and a half later. The day we were to be discharged, an ECHO showed moderate to severe cardiomyopathy. Alex was two weeks old.

During all that time, Alex hardly showed any signs of distress except breathing faster at the very beginning. I strongly believe he is still with us only because of the highly professional care and attention of all the staff we met along our way.

A week after the diagnosis of dilated cardiomyopathy and introduction of captopril, we were sent home. We arrived late, Alex was half asleep, and I couldn't believe he was with us in his own bed just next to ours. I have never been happier than that day!

In another couple of weeks, we went back for a repeat ECHO and were asked to run a Barth syndrome genetic test. That was the first time in my life that I had heard of the disease. Before our next review with the Cardiology consultant, I read everything I could find about the syndrome. The picture was not very pleasant. Understanding that my son has an unpredictable future was the worst bit. At this time, Alex was started on digoxin, furosemide and spironolactone. After the appointment, I contacted Annick Manton who warmly welcomed me into the Barth community.

Thanks to the support of several families, I feel more confident and ready to fight for my son. I can't express how valuable the time spent with them was and how helpful their advice was! I can't wait to meet all the rest of you at the next clinic!



Alex (age 5 months)
(Photo courtesy of Alex's family²⁰¹⁵)

The Barth Syndrome Foundation listserv and its archives are the best sources of information for me at the moment. There is always somebody at another computer who has been in your situation and can give you advice. Michaela Damin, Chair of the Barth Syndrome Trust, kindly introduced our family and guided us through the beginning of understanding life with Barth syndrome.

At the age of two months, Alex was seen at the Bristol Royal Hospital for Children and started on G-CSF as suggested by Professor Colin Steward. Nicol Clayton is overseeing Alex's diet and is still very involved in all his weight problems. Together they made my dream of bringing up my happy boy real.

Alex is nearly five months now and developing normally. Despite difficulties with feeding (his daily intake is unsurprisingly not great), his length and weight nearly follow his birth centile lines. All milestones have been achieved; actually he is even a bit advanced. G-CSF treatment is keeping him out of hospital and sufficient nutrition, now with a naso-gastric tube, along with cardiac medications, help him to thrive. Cardiomyopathy is mild to moderate, with no hospital admissions for the last month. He likes playing, meeting people, and he is very curious and active! I get more tired than he does by the end of the day!

Alex is a very happy boy, and we are so lucky to have him! I don't know what awaits us in the future, but we are definitely more prepared to fight it with the support of the Barth Syndrome Trust!

In The News

The Vavasour family who lost their son, Sebastian, tell their story on how an organ donor could have saved their son's life.



Laura & Sebastian

"... It was on the day that her husband returned to work after paternity leave that Laura Vavasour first realised that all was not well with their newborn son. Having been alert and feeding well, at 17 days old Sebastian was struggling to feed and was unusually hot. Laura — who was in her final year of training to be a GP — was assured by her doctor that he was fine, but her instinct told her otherwise. ..."

To read the full article please visit: <http://www.telegraph.co.uk/women/health/an-organ-donor-could-have-saved-our-son>

(Photo courtesy of Laura²⁰¹¹)

What's On My Mind

Barth Syndrome Foundation of Canada

By Susan Hone, President, Barth Syndrome Foundation of Canada

As I was packing for the BSFCa's annual planning meeting, I was thinking of how many things in my life are associated with Barth syndrome and the BSFCa, some good and some bad. November is the month Barth syndrome never leaves my mind for a day or even an hour; too many related anniversaries. Anniversaries are usually thought of as a reason to celebrate, although at times they are in fact a memory of an event that you wish never happened.

November 19, 1989 was the day my son, Jordan, received his new heart. He was two years old. It was like he was starting his life over. Everything was going great, and I was anticipating celebrating the first anniversary of his new heart, when instead, on November 15, 1990, Jordan died, four days short of his heart anniversary. It was in November 1990 that an article by Dr. Richard Kelley on Barth syndrome was published. That article would lead to the diagnosis of our son, Jared, born in 1994 with Barth syndrome and solve the mystery of Jordan's illness. On November 19, 1992 my unaffected son, Joshua, was born — the third anniversary of Jordan's transplant.

In November of 2004 I attended my first volunteer session for BSFCa. The planning meetings for BSFCa are typically held in November. It's our one time a year to get together and reflect on the previous years, remember those who have impacted on our lives, rejoice at our accomplishments, and renew our commitment to *Enhance the lives and outcomes of Canadian individuals and families affected by Barth syndrome.*

Before our planning session in 2014, I traveled with other members of BSFCa to attend a memorial service for a young man who had recently passed away from complications of Barth syndrome. That same year, I accepted the position of President of BSFCa.

As 2015 comes to an end and we plan for 2016, I look forward to continuing to work toward the day when everyone's Barth syndrome anniversaries are good ones.



Susan Hone
(Photo courtesy of Susan Hone²⁰¹⁵)

From the Heart

By Audrey Hintze, Grandparent of Affected Individual; Volunteer, Barth Syndrome Foundation of Canada

December 22nd — what a wonderful Christmas present. My daughter, Lynn, had just given birth to my first grandchild — a beautiful boy whom they named Adam. He was the sunshine that came to lighten my rather sad home at that time, and this, almost 26 years later, remains true, as he is a delight.

It wasn't long after that we received the news that Adam had a heart defect. My reaction was — Oh no, not again! I, along with my sister Phyllis, had both lost little boys, due to an unknown heart ailment. Years later, this condition had a name — Barth syndrome, and it launched us into a whole new world.

Adam showed such knowledge at a young age — I think he was just two when he told me that I had an agitator in my washing machine. At a very young age, he knew all the countries (including in Africa) and their capitals, as well as all the kings and queens of England.

He has an inquisitive mind and loves mechanical things. He is a whiz at repairing small motors, generators, snowblowers, etc. He loves the challenge. We proudly attended his college graduation.



Audrey & Adam
(Photo courtesy of BSFCa²⁰¹⁴)

(Cont'd on page 22)

From the Heart

(Cont'd from page 21)

He also loves gardening, and each year he helps me with the cleanup in my large garden. He helped build a deck for me and has power-washed it several times. I am happy to see him continue this passion in his new home — helping his parents put in a new garden.

We all look forward to the biennial Barth Conference held in Florida. The researchers and medical personnel bring us up-to-date on the latest treatments, and it is a time to renew friendships. Adam has the wonderful opportunity of meeting up with other Barth men/boys who have so much in common. They each know what the other ones are experiencing — the daily frustrations — and he eagerly awaits the next conference in July 2016.

I have had the pleasure over the years of raising funds for much-needed research. It is my sincere hope that others who are in, or approaching their golden years, will remember the Barth Syndrome Foundation of Canada or any affiliate in their final wishes. Hopefully, we can celebrate a medical breakthrough that will help all of the world-wide boys/men, like Adam, who are waiting and praying for that day.



(L-R) Robert (age 29), Travis (age 20), Ryan (age 24), and Adam (age 25) (Photo courtesy of BSFCa²⁰¹³)

Let's Have a Party!

By Cathy Ritter, Board Director, Barth Syndrome Foundation of Canada

One of the quintessential must-dos during the summer is to have a party! With that “spirit” in mind, the Summer Party Starter Kit was invented and some lucky winner (or two!) would receive the beverages required for a grand summer party. With the generous donations of many BSFCa supporters, two large tubs were filled with all kinds of wine, liqueurs and alcohol to raffle off in support of the Barth Syndrome Foundation of Canada. Those same supporters then aided in the selling of the raffle tickets so that almost \$3,000 dollars was raised and donated to the Foundation. Amazing!

On June 28th, Ryan drew the winning tickets. Michael Hope, very well known in the Barth community, was the first prize winner, while Phil Bourget RN, was the winner of the second prize. As an added bonus, each of the baskets contained one of the popular Barth Bears! Congratulations to the winners.

A big THANK YOU also goes out to all of the individuals who helped make this a success: the people who donated the prizes, sold tickets and, of course, those of you who bought them! A message to those who were disappointed that they didn't win... you will get another chance in the summer of 2016!



Ryan draws the winning raffle tickets for the Summer Party Starter Kit



Cathy Ritter presents the runner up of the Summer Party Starter Kit, Phil Bourget, with his prize (Photos courtesy of BSFCa²⁰¹⁵)

A Family Unites for Fun and Barth

By Lynn Elwood, Board Director, Barth Syndrome Foundation of Canada

Our “Wonderful Wooden” raffle this fall was a great way to inspire our family to band together for Barth!

We recruited all ages. Justin (Adam’s brother) and his girlfriend, Dalaina, spent many hours proofing, collating, and numbering raffle tickets that were designed by Lynn and Rick. Adam helped with ticket pick-up and deliveries as well as trips to Lee Valley for clock parts.

The draw prizes are truly handmade by family. Dad (Les) planed the rough pine lumber, selecting just the right boards and then hand crafting the three nesting tables and the pine wall clock with its beveled glass dome. Lois helped as well, especially in the finishing stages.

Adam lovingly and gently turned a stick of butternut wood on the lathe into the base for a ball-point pen. He then hand assembled, waxed and polished his finished product. The resulting prizes are beautiful, professional items that were made with love and pride, and will be special additions to the winners’ households.

Many people have shared in the ticket sales. Lynn recruited her Mom, Audrey, and Aunt Phyl to sell tickets to friends, and Lois asked the ever dedicated BSFCa volunteers, Carol Wilks (Adam’s Aunt) and other friends to sell tickets as well. We also have four very Barth family friendly folks from Florida (Sharon, Jan, Lee, and Joanie) selling tickets to friends. Any US based winners can expect a January delivery!!

This activity has been great fun, and we all anxiously await the draw date on Sunday, November 22nd! Thank you family members, friends, volunteers, BSFCa executive members and huge hugs to the MANY, MANY TICKET BUYERS!!! You have all shared in our dream and vision of *“enhancing the lives and outcomes of Canadian individuals and families affected by Barth syndrome.”*



Wonderful wooden raffle prizes
(Photos courtesy of BSFCa 2015)

A New Direction

By Chris Hope, Secretary/Treasurer, Barth Syndrome Foundation of Canada

This year, Barth Syndrome Foundation of Canada (BSFCa) has taken a different fundraising route. In the past, we held one large yearly event—the annual Barth Syndrome Foundation of Canada Driving for the Cure Golf Classic. After ten years of extremely successful tournaments, we figured it was time to go in a new direction. Although as a corporation we still have our annual appeal (this year boosted by a generous \$5,000.00 matching donation) our remaining fundraisers have been individual campaigns run by family and friends on behalf of BSFCa. We are very grateful for all of the time and effort everyone puts into these events and personal appeals. As the year is coming to a close, we have another matching challenge for the month of December to help us out, and we continue to search for grants for which to apply.

With multiple individual fundraisers and special contributions, we are happy to say that although we missed seeing all our golfing friends, we have had a successful year financially, and we are in great shape as we start to make our plans for 2016.

Association Barth France

From a Marathon Runner's Point of View...

By Florence Mannes, Chair, Association Barth France



Raphaël (age 7) & Xavier,
member of Team Barth France
Deauville Ironman - June 2015

My very first contact with the Barth Syndrome Foundation (BSF) was just before we received a diagnosis for our son, Raphaël. Barth syndrome was suspected, but we were still waiting for the genetic confirmation. As we had no idea of what this would mean to us, I searched “Barth syndrome” on the internet. This was in August 2009 when our son was nine months old. Although I was devastated by what I was reading, I couldn’t help reading, and reading again, all the articles, pages, and educational information available on the Barth Syndrome Foundation’s website. I wanted to know as much as I could about this disease that was possibly affecting our boy.

Once we got the diagnosis, I went back to the website, and read again. It took me several weeks before I dared to send an email to Shelley Bowen, Director of Family Services (BSF) to introduce ourselves.

The worst part of reading all those articles was that, at that time, though there were many doctors and researchers working on Barth syndrome, we had the feeling that these doctors were still trying to get a better understanding of the disease, rather than searching for a cure.

As a marathon runner, it felt like I had decided to run the most difficult marathon ever, not knowing at all how I would train for this, and not knowing either if I could one day run this marathon.

One year later, we attended our first Barth Syndrome International Scientific, Medical & Family Conference. It was amazing to see all these families, doctors, and volunteers working together to achieve the same goal: finding a cure for our boys.

Now, we knew that we had a team to rely on, with which we would run the marathon. We were not alone anymore...but we still had no idea of how and when we would run this marathon.

As the years went on, more and more researchers started to really work on finding a cure, not only on understanding the disease...

We were running the marathon, with a huge and dedicated team, but it is such a long road...and we were so terribly sad that some of our boys had to quit the race before the finish line, before a cure was found.

(Cont'd on page 25)



Team Barth France at the Paris Bike and Run

(From left to right): Philippe, Romain, Victor, Cédric, Florence, Philippe, Audrey, Gilles, Stéphanie, Laura & Martine

From a Marathon Runner's Point of View...

(Cont'd from page 24)

Every year, especially when we read the research grants that have been awarded by BSF and its affiliates, we have the feeling that things are going forward, that we are at the edge of new therapies...from ideas of a cure to testing a cure. The road is long, sooooo long; hard, sooooo hard...but we need to keep hope alive and to keep going forward.

If we want to succeed, if we want to cross the finish line of this “Barth marathon”, we have to keep going, to fight, to find funds to support the research. The marathon for finding a cure for Barth syndrome is a long journey, but we are now experienced runners, aware of how difficult this challenge is and so willing to cross the finish line. We have all the capability and the ability to do so...and I am convinced we will succeed, for our boys who are still fighting, and for those whom we have lost. My hope is that one day our boys will be able to run a 26.2 mile marathon, once the “Barth marathon” is behind them.

Barth France Traditional Fundraising Events

During the last six months of 2015, Barth France has raised funds through many events that have now become traditional for the Association:

Multiple Races

- Marseille-Cassis
- Paris Marathon
- Course des Héros
- NYC Marathon

Multiple Triathlons

- Ironman France Nice
- Embrun Ironman
- Vichy Half Ironman and Ironman
- Deauville Triathlon
- Roth Ironman

Multiple Grassroots Fundraisers

- 5th Golf Tournament (photo)
- 3rd Gospel Concert
- 4th Poker Tournament
- 2nd Charity Dinner (Black Truffle)



Barth syndrome hits the news



4th Barth France Poker Tournament in Paris
(Photos courtesy of Barth France²⁰¹⁵)

Proud to Run for Barth

By Didier Brignant, Member of Team Barth France



Team Barth France at the NYC Marathon
(including Corinne Levezuel, Didier Brignant & their friends)

The adventure started in our apartment in Nice more than a year ago. The starting point was my forthcoming 40th birthday. What would be the most remarkable gift? The most outstanding one? New York, of course! The New York City Marathon. From there, everything went very fast. Emails were sent, relatives were called, and friends were enrolled into what would be a memorable event.

Then came the obvious. Not only were we about to experience an amazing human adventure, we also had to run for an altruistic purpose. I immediately thought about Florence and her commitment to Barth France. Well... let's go for it! We will run for Raphaël and all the children who fight without respite against this unknown disease.

(Cont'd on page 26)

Proud to Run for Barth

(Cont'd from page 25)

Through the social networks, we told our friends and tried to raise awareness. Informational meetings, distribution of leaflets... things needed to be done in order to communicate about the disease. The press was contacted and agreed to participate. Nice Matin (French Local Daily), Direct Matin (French National Daily) and France Bleu Azur (radio station), published articles about us. All the while, training sessions continued three times a week. We may have suffered sometimes, but we knew it would be a beautiful race for a beautiful cause. The event webpage was created, and 550€ was raised (US \$585.50).



Finally, it was time to fly to New York. We packed our Barth T-shirts and our flag ready to cross the finish line. On D-day, we didn't forget the group photo, or shall I say, the family photo. **We are all Barth.**

Our team consisted of nine runners, supported by two million people in the streets. It was a beautiful day. New York is definitely magic. After finishing in 4h15, I was happy and proud to have run for Barth.

Jules' Pretty Normal Life

By Valérie Lallemand, Mother of Jules (age 6 1/2), Belgium

We are one of the two diagnosed French speaking families living in Belgium. We live near the city of Liège in Wallonia, which means we are closer to Germany and The Netherlands than to the seaside or even France!



Jules
(Photo courtesy of Jules' family)

Our son, Jules, was diagnosed when he was three months old. However, complications were present when he was three days old when the doctors discovered that his heart worked poorly. After a month in hospital, we finally were able to go home. When Jules was around two months old, his heart was doing better and function was within the normal range again. However, he was eating very poorly and needed a feeding tube when he was three months old. He is now six and a half years old, he is still tube fed, and he is doing fine. Of course he gets the regular medical appointments Barth syndrome boys are used to, to check his heart, the feeding issues, and the neutropenia.

has a swimming lesson every week and likes it a lot but likes playing in the pool afterwards even more. He tried pony riding last year but did not like it when the pony trotted too fast. He loves watching television and especially enjoys the Harry Potter and Narnia Chronicles movies. He then replays the scenes with his Playmobils. This summer, he adopted a little red cat. He called him Harry (of course) and loves him very much.

So, as you can see, Jules has a pretty normal life, nearly the same as the other children of his age except for the regular appointments with the doctors and the daily medication for his heart, and neutropenia. We are also more vigilant about his health and visit the ER more frequently than others when he gets a fever.

He started primary school in September. That meant a change of school and rhythm. He used to come back home for lunch every day while in Kindergarten and now stays at school where a nurse comes to feed him there at lunchtime. Learning to read seems to be a challenge right now and he prefers to play with his Playmobils.

Jules is a very curious and active little boy. He likes to discover new things. He always needs to do something and can't stand still and always needs to move. Playing outside in the garden is a must. He

Being in contact with the Barth Syndrome Foundation, the Barth Syndrome Trust and the NHS Service in Bristol, and Barth France is very important to us. We gather precious information from this community who always has an understanding ear to listen to our problems.

Last January, we also attended the first French meeting in Paris. That was a great opportunity to meet other French speaking families, the doctors and researchers. Exchanging information is a life saver!

Barth Italia's Historic Meeting

By Paola Cazzaniga, President, Association Barth Italia

On September 30, 2015, we had our first meeting of Barth Italia, the Italian Barth Syndrome Families Association. The meeting was a satellite of the Workshop "Cardiolipin — A Key Lipid of Mitochondria in Health and Disease - 2nd Edition" held in Florence (Italy) on September 30 and October 1, 2015, which was co-organized by Dr. Angela Corcelli (Department of Basic Medical Sciences, Neurosciences and Sensory Organs, University of Bari A. Moro, Bari, Italy) and Dr. Michael Schlame (Departments of Anesthesiology and Cell Biology, NYU Langone Medical Center, New York, NY, USA).

Five of the seven families diagnosed in Italy were present. Some of the parents met for the first time, and this was their initial opportunity to share experiences. Also, two boys, Pietro and Valerio, met for the first time! Two small children, Mattia and Ruben, were there and their cheerfulness was a pleasure for everybody to see. Pietro, 13 years old, and the oldest of the boys, described his life, his difficulties, and how he gets along with the help of his family and of his many friends. Pietro was very nervous and emotional, but he gave a great talk!

Among those present were Dr. Alice Donati from the Meyer Hospital in Florence, Dr. Gasperini from the San Gerardo Hospital in Monza, and Dr. Stoppa (pediatric neuropsychiatrist). Dr. Alice Donati has been for many years, and still is, the reference point for the disease in Italy. Dr. Gasperini's expertise is in metabolic disorders. These specialists described from their different points of view the extreme variability of the disorder and highlighted the possible needs and the difficulties for patients and families during the different life periods. They also outlined the need for physiotherapy and school support.

Prof. Colin Steward from the Bristol Clinic in the UK was an active part of the discussion. He gave the families the opportunity to describe their experiences and shared his vast clinical experience with Barth syndrome families in the UK. His ideas and suggestions for the hematological aspects of the disease gave rise to a very intense discussion and many questions.



Families gather at first meeting of Barth Italia



(L-R) Dr. Michael Schlame, Dr. Daniela Toniolo & Dr. Colin Steward

Finally, Dr. Amelia Morrone, the geneticist at the Meyer Hospital, presented a detailed description of problems and advances in diagnosis. Considering the young age of some of the mothers and at least one carrier sister, we think it was very relevant to have clear information of this very delicate topic.

At the end of the meeting, the families and some of the clinicians and experts had an informal lunch together. It was great to be able to share goals of Barth Italia, ideas on how to raise funds, and the emotion of being together for this very demanding challenge of our life.

At the end, we have to thank the scientific committee of Barth Italia, Prof. Colin Steward, for coming all the way to Florence, and all the other friends who supported our efforts. We have to give a special thanks to Prof. Angela Corelli for organizing the workshop and supporting the organization of the first meeting of Barth Italia from many points of view. We would also like to thank the Barth Syndrome Foundation for financially supporting this important meeting.



(L-R) Ruben (age 14 months) and Valerio (age 11) with their dads, Diego & Carlo (Photos courtesy of Barth Italia²⁰¹⁵)



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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**



Ty (age 1)
(Photo courtesy of Ty's family)



Bob (age 55)
(Photo courtesy of Bob's family)