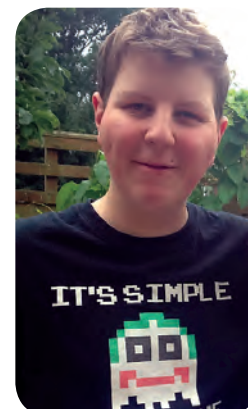


Helping to Find a Treatment for Everyone

By Nicholas, Affected Individual (age 18), United Kingdom

"I volunteer for Barth syndrome research as often as I can. Because if I don't help, if we all don't help, then how can there be progress? ... I do the studies, not just for me; I do the studies to find a treatment for everyone." ~ Nicholas, Affected Individual (age 18), United Kingdom

When I was in the United States in the summer of 2016, I volunteered for Dr. Todd Cade's exercise study, "Heart and Skeletal Muscle Metabolism, Energetics and Function in Barth Syndrome". Todd's study wasn't always comfortable or easy, and a lot of it was boring, but it's worth putting yourself through that for just a day or two to try to find a treatment because studies like this are the best way of finding out more about Barth syndrome and finding better ways to judge whether new medical therapies are being effective.



Nicholas (age 18)
(Photo courtesy of Michaela 2016)

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I volunteer for Barth syndrome research as often as I can. Because if I don't help, if we all don't help, then how can there be progress? We're the only test subjects — it's not like cancer research where millions of people are affected. There are just 200 of us in the world and if we don't do the studies, do the tests, there will be no treatments and no cure. The only people that scientists or researchers can test things on are us.

Growing up, I always knew that I had Barth syndrome, and that I had to live with it, but I never really knew what it was until years later. I had lots
(Cont'd on page 7)

2016 Research Grant Cycle Awardees

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



Dr. Matt Toth
(Photo courtesy of Amanda Clark 2016)

The 2016 cycle of the Barth Syndrome Foundation (BSF) Research Grant Program marks the 15th year of this highly successful plan to increase our understanding of Barth syndrome so that specific therapies may be developed. Many of the therapies that were discussed in detail at the 2016 Barth Syndrome International Scientific, Medical & Family Conference have their origins or have been supported by this BSF program. Bezafibrate therapy, gene therapy, enzyme replacement therapy, exercise therapy, and lipid replacement therapy can all be tracked to the support that BSF research grants have provided over the years. We hope to continue this tradition with the 2016 awardees.

(Cont'd on page 4)

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BSF in Beantown

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

You may not realize this, but the Barth Syndrome Foundation (BSF) is a completely virtual organization. We have no physical headquarters. All four of the full-time staff members work from home offices, as do our part-time contractors. Our staff and volunteer board of directors are scattered throughout the United States and abroad. That makes coming together in-person a rare treat, something that happens only once or twice per year.

Our recent face-to-face board meeting in the Boston area offered a great opportunity to discuss several important topics in our two days together. Susan McCormack hosted the board of directors and staff in her home for the meeting.

We discussed finance, fundraising, science and medicine, family services, awareness, and revising our strategic plan. We also tackled the 2018 conference. Deciding on the location requires months of research on all available options. We look for low rates, a family-friendly location, access to an international airport, proximity to top-notch hospitals, affordable restaurants in walking distance, and much more. After very thorough and careful consideration, the board voted to sign a contract with the Hilton Clearwater Beach Hotel for the 2018 conference.

I'm excited to share that the hotel has undergone some significant renovations since they hosted us in 2016, including: two new, zero-entry pools and pool decks, a completely new lobby with lobby bar, a Starbucks coffee shop, and a new, Mexican restaurant! We are very excited to go back to the Hilton Clearwater Beach for our 2018 conference. Be sure to mark your calendars for July 16 – 20, 2018!

The board appointed Susan McCormack as BSF's vice chair. Susan will take on the role of chair in April, 2018 when Marc Sernel's term ends. This will give Susan a full year to "shadow" Marc in the role before assuming responsibility. Kevin Woodward was named treasurer, as Randy Buddemeyer's term ended in April, 2017. We wish Kevin luck in his new role, and we give thanks to Randy for his years of service on the board!

BSF remains on solid footing as a small but mighty organization. Our board of directors and staff work hard, along with our volunteers and donors, to fulfill our mission — *saving lives through education, advances in treatment, and finding a cure for Barth syndrome*. No one of us can do it alone. What an exciting time to be part of this journey together; thank you for your support along the way!

Lindsay B. Groff

Lindsay B. Groff
Executive Director



BSF board members and staff (Photo courtesy of BSF 2017)

Reaching Our Full Potential

By Marc Sernel, Chairman, Barth Syndrome Foundation

All you can ever ask of someone is for them to strive to reach their full potential. A person's "God-given potential" is inherent in who they are, and factors outside of their control can make achievement more difficult for some than others. We can only try to do the best we can with the hand we are dealt.

All of us that are parents dream for our children, hoping they can grow up to be successful and happy adults. There is no one-size-fits-all approach to parenting. Among the spectrum of parents in the world, it's probably safe to say that I'm more demanding than some, setting high expectations for my children and then encouraging them to work to achieve those expectations. Parenting a child with Barth syndrome is more difficult, on many levels. Aside from the obvious medical challenges and crises, there is also the difficulty of knowing how much you can or should "push" a child that you know has physical limitations that other children do not. Finding the right balance is not easy. But I want my son with Barth syndrome to know that I still expect him to do his best, and that I have big dreams for him just as he should have for himself.

Similar to our Barth children facing and overcoming challenges to live their lives to the fullest they can, the Barth Syndrome Foundation strives to achieve the most with what we have. Like our Barth boys and men, we try not to dwell on our limitations or confine our goals to what presently seems attainable. We try to focus on what we can do, not what we can't do. And we don't stop or turn around when confronted with obstacles or roadblocks on the way to our goals; we find ways to go around, over or through the things that stand in our way. The amazing spirit and can-do attitude of our Barth boys and men guides our organization and its never-ending quest to reach our goals.

We are an organization that is relatively small in numbers and resources. Even among our peers in the rare disease community, we are considered tiny, ultra-small even among the "ultra-rare" diseases. But as the saying goes, it's not the size of the dog in the fight, but the size of the fight in the dog. We may be small, but we have some "fight" in this organization, people that don't take no for an answer and have a can-do approach to life. This organization would not exist, and many of our boys would not have been diagnosed with Barth syndrome or received the tailored care and treatment that they have, without the tireless efforts of parents that simply would not give up.

This organization is only as great as we make it. And by "we" I mean everyone reading this newsletter, our entire Barth family worldwide. The progress we've made since 2000 is truly remarkable, and it's important to recognize the important milestones we've achieved. Blood, sweat, and tears have been shed—literally—to achieve the many successes we have had to date. These successes would not have been possible without the contributions of many, ranging from the extraordinary efforts of our volunteers to the millions of dollars raised by the incredible fundraising efforts and generosity of our donors. But there's a lot more work to do. More blood will be needed, in the form of participation in clinical trials and research that is ongoing and to come in the future. More sweat will need to be poured by our BSF volunteers, including the many caring doctors and scientists that are on this journey with us. More tears will flow as we continue to face the cruel challenges and heartbreak that Barth syndrome presents. And we'll need to raise millions more in donations in the coming years to continue to push the science forward to achieve the goal of a treatment and ultimate cure for Barth syndrome.

No single person, family, or subset of this group will be able to make this happen. We need everyone to contribute what they can for BSF to reach its full potential, and we look forward to both new and longtime members signing on for new roles and responsibilities. This past April marked the end of Randy Buddemeyer's tenure on the board, and we thank Randy for his long-time dedication and service to our organization. While we'll miss Randy's laser-focus and financial acumen as our treasurer, I look forward to working with our new treasurer, Kevin Woodward, and our talented board and staff to carry on our mission. With others stepping up and all pitching in, we will push this organization to its fullest potential and reach our goals together.

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2016 Research Grant Cycle Awardees

(Cont'd from page 1)

| | |
|---|--|
| <p>Abbreviations and definitions: AAV — adeno-associated virus (a vector commonly used in gene therapy) CL — cardiolipin (the lipid altered in Barth syndrome) MLCL — monolysocardiolipin (altered cardiolipin missing one fatty acid chain) KD — knockdown mouse model of Barth syndrome <i>tafazzin</i> — the gene that codes for the protein that when defective causes Barth syndrome iPS cell — induced pluripotent stem cell lines ROS — reactive oxygen species (too much of this compound may be pathological)</p> | |
|  | <p>Christina Pacak, PhD, Assistant Professor, University of Florida, Gainesville, Florida</p> <p>“Optimization of AAV-mediated gene therapy for Barth syndrome”</p> <p>Award: US \$100,000 over 2-year period</p> <p><i>*Funding for this award was provided by the Will McCurdy Fund for Advancement in Therapies for Barth Syndrome and Barth Syndrome Foundation of Canada</i></p> |
| <p>Using iPS cell lines to “fine-tune” the gene therapy vectors designed for treating Barth syndrome. Gene therapy is a novel procedure that has the potential to be curative for Barth syndrome and for other genetic diseases. Because of its complexity and the many unknowns of this new medical technology, it is prudent to optimize the performance of the actual delivery vectors or viruses before starting clinical trials. To minimize cost and potential side effects, Dr. Pacak’s optimization studies will answer questions like which vector transduces cells (introduces the normal <i>tafazzin</i> gene) most efficiently and delivers functionality as measured by MLCL/CL ratios, decreased ROS production, etc. Dr. Pacak’s project will use iPS cells isolated from about two dozen Barth syndrome individuals to test the current AAV vectors for efficacy and to “fine-tune” the expression of the <i>tafazzin</i> gene. She will optimize these AAV vectors by screening for the best promoters to drive the transduced <i>tafazzin</i> gene’s expression and by screening for the best human codons that can be easily translated into fully functional <i>tafazzin</i> protein. Dr. Pacak has demonstrated that the current AAV vectors do indeed reverse the skeletal muscle dysfunction and lower the heart mass of the KD mouse model, and they correct the MLCL/CL ratio in Barth syndrome iPS cell lines. She will use the results of this proposed study to further improve the chances of success in future gene therapy clinical trials.</p> | |
|  | <p>Deborah Tribouillard-Tanvier, PhD, Permanent Researcher, CNRS, University of Bordeaux, Bordeaux, France</p> <p>“Discovery of drug candidates for the Barth syndrome using a yeast-based screening approach and higher eukaryotic models of this disease”</p> <p>Award: US \$44,000 over 2-year period</p> <p><i>*Funding for this award was provided by Association Barth France</i></p> |
| <p>Further testing of over 20 compounds from a collection of FDA-approved drugs identified in a yeast-based screening experiment. Dr. Tribouillard-Tanvier and colleagues used a novel yeast-based screen (where the <i>tafazzin</i> gene was deleted) to identify over 20 compounds in the Prestwick collection of 1,280 FDA-approved compounds (drugs) that might offer potential treatments to Barth syndrome. They propose to expand the testing of these compounds by using cellular models of Barth syndrome and eventually the knockout mouse model being developed by Dr. Strathdee, who is one of the collaborators. Preliminary data has validated the yeast screen by showing genetic suppression of the screening yeast cells by overexpression of a mitochondrial membrane protein. Dr. Tribouillard-Tanvier will use: a HeLa cellular model of Barth syndrome, lymphoblastoid cell lines from Barth syndrome individuals, and iPS cell lines derived from Barth syndrome individuals to determine how these compounds are acting and if they are worthy to proceed to the next level of testing. The next step will be to use the knockout mouse model to test for compound efficacy. By working with compounds from the Prestwick collection, further development of positive results should accelerate any drug approval process.</p> | |

2016 Research Grant Cycle Awardees

(Cont'd from page 4)

| | |
|---|--|
|  | <p>W. Todd Cade, PT, PhD, Professor, Washington University, St. Louis, Missouri</p> <p>“Characterization of the ‘metabolic phenotype’ in Barth syndrome with cardiac transplantation”</p> <p>Award: US \$49,820 over 1-year period with \$21,000 in travel expenses</p> <p><i>*Funding for this award was provided by the Will McCurdy Fund for Advancement in Therapies for Barth Syndrome</i></p> |
| <p>Does cardiac transplantation alter the unusual metabolism of Barth syndrome individuals? Dr. Cade has defined the unusual metabolism of Barth syndrome individuals by performing several clinical studies. In brief, Barth syndrome individuals metabolize or “burn” the sugar called glucose to an inordinate degree to obtain energy for living, and they reduce or downshift the “burning” of fatty acids which are the predominate energy sources for the heart and skeletal muscles when the individual is at rest. To briefly summarize, Barth syndrome individuals are not efficient in their use of energy sources (fats, carbohydrates, proteins). It is believed this inefficiency in utilizing different energy sources explains the fatigue of Barth syndrome. In all the Barth syndrome clinical studies so far, individuals with transplanted hearts were excluded for scientific reasons. Dr. Cade would like to determine the characteristics of this cohort of heart-transplanted Barth syndrome individuals (15% to 20% of all of them) and compare them to what he has found in his other studies. Dr. Cade’s work (with 29 individuals) has shown that in addition to their low utilization of fatty acids during exercise, Barth syndrome individuals do not metabolize or “burn” amino acids to the extent originally thought to be pathological, but rather they show an extraordinarily high metabolism of glucose or sugar at times of rest, at exercise, and during recovery after exercise. Dr. Cade will compare what he knows from his work with the non-transplanted group of Barth syndrome individuals to the heart-transplanted group by observing for any metabolic or symptom differences. Studying this unique subgroup could provide us with a better understanding of the long-term effects of Barth syndrome both in the heart-transplanted and the non-transplanted groups.</p> | |
|  | <p>Shanta Dhar, PhD, Associate Professor, University of Miami Miller School Medicine, Miami, Florida</p> <p>“Cardiolipin enhancing biodegradable nanoparticle for Barth syndrome”</p> <p>Award: US \$50,000 over 2-year period</p> <p><i>*Funding for this award was provided by the Science & Medicine Fund</i></p> |
| <p>Using nanoparticles as a lipid replacement therapy for BTHS. Dr. Dhar will use her expertise in constructing small particles, nanoparticles, to make varieties containing cardiolipin and coenzyme Q10 as a potential therapy for Barth syndrome. These nanoparticles may be able to replace the cardiolipin that is missing in the mitochondria of Barth syndrome individuals. Mitochondria are the parts of the cell that provide most of the energy needed to live. Furthermore, the addition of coenzyme Q10 to the nanoparticles may also help to reduce the dysfunction associated with Barth syndrome mitochondria. After making these nanoparticles, Dr. Dhar will test them on models of Barth syndrome including the heart-on-a-chip system from iPS cells developed by Dr. Bill Pu and colleagues and the KD mouse model. Dr. Dhar has made cardiolipin containing nanoparticles and shown that they can fuse to mitochondria and are not toxic to the cells. She will also analyze these CL nanoparticles for toxicity.</p> | |

BSF is truly fortunate to have dedicated researchers, like these 2016 awardees, interested in studying Barth syndrome. BSF highly values the effort and the time these professionals devote to our cause. The seed funding provided by these awards has provided the basis not only for generating therapeutic ideas, but has also supported these researchers to discover data needed to obtain larger grants from the National Institutes of Health (NIH) and other large-grant-giving organizations. So far nine prestigious R01 grants have been won by BSF researchers. These and other non-BSF grants multiply by a factor of five the dollars BSF has invested in its grant program. We expect that the 2016 cycle awardees will continue to add to this record of achievement in science excellence and clinical advancement.

A complete list of all grant awardees can be found on BSF’s website at www.barthsyndrome.org. (Photos courtesy of grant recipients ²⁰¹⁶)

Barth Syndrome Foundation

Research Grant Program

2017 Request for Research Proposals

The Barth Syndrome Foundation, Inc. (BSF) and its international affiliates announce the availability of funding for basic science and clinical research on the natural history, biochemical basis, and treatment of Barth syndrome. There are two categories: IDEA grants for 1-2 years and DEVELOPMENT grants for 2-3 years with budgetary maximums of US \$50,000 or \$100,000, respectively. BSF's Research Grant Program allows young, non-tenured investigators to include in their submitted budget up to 75% of the total grant amount as Principal Investigator (PI) salary. In addition, for clinical applications requiring that volunteers travel to a clinical research site, these travel expenses will be handled separately and excluded from the budget limitation. We encourage all investigators at every professional level to submit their best ideas. There are no geographical limitations to this funding.

Background

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

Types of Proposals Sought

We are interested in providing “seed grant funding” to young investigators as well as attracting experienced investigators new to the field of BTHS basic science or clinical research. We anticipate that these funds will be used for the testing of initial hypotheses and the collection of preliminary data leading to successful long-term funding by the National Institutes of Health (NIH) and other major granting institutions around the world.

Process

BSF has a competitive grant process. Applications should be of 10–15 pages in length and must follow the instructions listed on the BSF website. In general terms, detailed information about the specific aims, significance, research design and methods, personnel, facilities, and budget will be required. A one-page, “Letter of Intent” is required for DEVELOPMENT grant applicants, and has a due date of **September 1, 2017**. The “Letter of Intent” is optional for IDEA grant applicants. We strongly encourage the submission of letters of intent before the due date to allow ample time.

Completed applications (and/or “letters of intent”) will be forwarded to the BSF Scientific and Medical Advisory Board (as well as to expert outside reviewers) for confidential evaluation. Response to the “Letter of Intent” will be communicated within two weeks of receipt. Based on the recommendations of the BSF Scientific and Medical Advisory Board, the BSF Board of Directors will make the final funding decisions about the grant applications. Once the final funding decisions are made, BSF affiliates will decide which, if any, of the approved grants they would like to fund. Please review our “Grants Awarded” webpage for a listing of grants that BSF and its affiliates have awarded to date.

Funding

We anticipate awarding several IDEA and DEVELOPMENT grants each year. Funds will be available soon after the successful grant applicants have been notified in early March, 2018.

Deadline

The deadline for submission of the completed research grant application is **October 31, 2017**, and grants will be awarded in early March, 2018. The deadline for the one-page “Letter of Intent,” if applicable, is **September 1, 2017**.

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Helping to Find a Treatment for Everyone

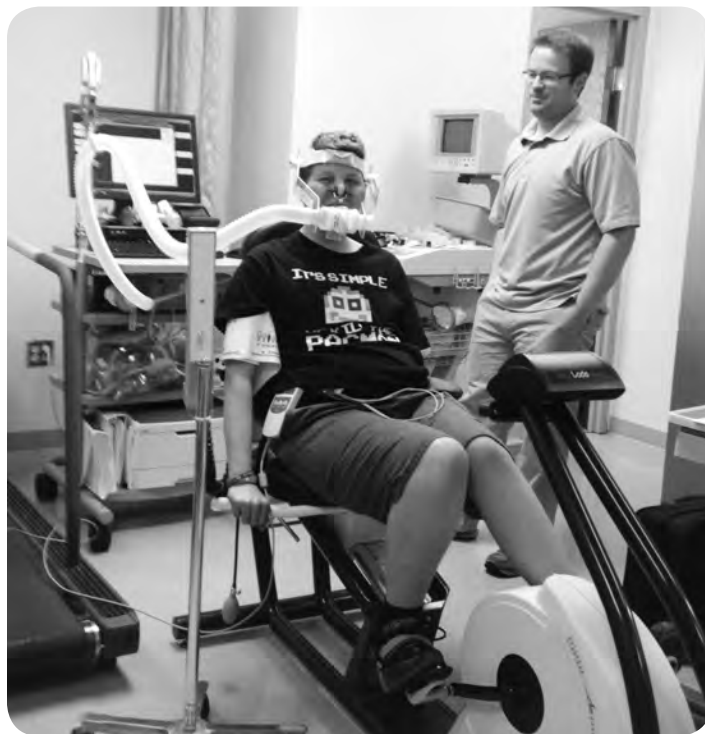
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of hospital visits and doctors' appointments and had lots of blood tests and took a lot of medications, but I just did it because my mom told me to because it was good for me. Parents will always look after you and try to do the best they can for you. But when it comes to taking part in research studies, your parents can't tell you to do it or not — they can advise you, but the choice is yours.

If I hear about a study from my mom or from Dr. Colin Steward or someone else who I know is trustworthy, then I am more likely to think about taking part in it because I know that they will have done plenty of checks to make sure that it's as safe as it can be. But that's not enough. In every clinical trial or medical study, there are always going to be some risks, some of which will be very unlikely and others more likely. We can't know until we read the information for ourselves. There's always a sheet that participants have to read which lists the pros and cons, and there are always people to ask if something is unclear. I've been tempted to skip this on occasion because reading it may be annoying or a little boring, but this is not a good idea! A single huge block of medical text makes me want to run away, and I think it might be easier for some of us if we could hear short podcasts through our Facebook page as another way of getting the information in smaller chunks that are easier to process.

When I was at the BSF conference in Clearwater in July 2016, I connected immediately with everyone. Not only to the Barth guys, but also to the siblings and parents. Everyone there just sort of knows. Back at home, I have some very special friends and family who accept me as I am. Walking with them means I don't fall behind or have to speed myself up because they all adjust to slow themselves down to my pace. But going to the conference and seeing everyone else was different. The guys there didn't have to slow down for me; they were already walking at my pace. So we could walk together. There would be a whole bunch of us affected guys and sibs sitting in the lobby, and we would have to decide whether to go to the pool or the sea, both of which are very nice. So we take a quick poll and without fail, every single guy with Barth syndrome votes for the pool, myself included. I ask: 'Why do you guys all choose the pool?' knowing that the reason I choose it is because I find walking in the soft sand too tiring. Sure enough, the answer was the same for each Barth guy. The best part was the blank expression on the sibs' faces — they didn't all get it, but we guys all understood that about each other.

Seeing everyone else having to go through the same things I go through creates such a strong connection between such a small group of people. I don't make a habit of feeling sorry for myself, but I feel for some of the guys who are not doing as well as I am. And, so I do the studies, not just for me; I do the studies to find a treatment for everyone.



Nick participates in Dr. Todd Cade's exercise study
(Photos courtesy of Michaela 2016)

What Does it Smell Like?

By Angela Corcelli, PhD, University of Bari Aldo Moro, Bari, Italy

This was one of the questions submitted to Barth syndrome patients who took part with their families at the 2016 Barth Syndrome International Scientific, Medical & Family Conference in Clearwater Beach, Florida. On this occasion, Italian scientists from the University of Bari carried out a psychophysical test in order to investigate olfactory sensing ability in Barth syndrome. Olfaction has different important functions that are usually underestimated in modern life. For example, it plays a critical role in the formation of individual food preferences and of healthy food habits in early life; an alteration of the sense of smell may lead to feeding problems, even causing vitamin deficiencies. Regarding Barth syndrome boys, parents and caregivers often report peculiar food preferences whose cause is not clear yet. The children seem to prefer strong flavors, suggesting a potential deficit in flavor perception; the question arises whether their olfaction might be altered too. So, a better understanding of how olfaction and taste works in Barth syndrome subjects could be useful to design a correct diet to provide needed calories and vitamins for those affected.



Drs. Angela Corcelli, Michele Dibattista
& Simona Lobasso
(Photo courtesy of BSF 2016)

The clinical trial, funded by the University of Bari and approved by its Ethical Committee, was based on the Sniffin' Sticks Test developed by Professor Hummel from the University of Dresden in 1997 and since then widely used to study human olfactory ability. "Sniffin' Sticks" are pen-like odor dispensing devices useful to test nasal chemosensory performance. Pens are filled with an odorant; removal of the cap will release the odor. The complete test consists of three sub-tests of olfactory function, namely tests for odor threshold ("rose" odor testing by means of a staircase with forced choice), odor discrimination (16 pairs of odorants, triple forced choice) and odor identification (16 common odorants, multiple forced choice from four visual items per test odorant).

On their arrival, the Italian team had a meeting with Shelley Bowen (Director, Barth Syndrome Foundation [BSF] Family Services & Awareness) and Michaela Damin (Chair, Barth Syndrome Trust), to schedule several trial sessions in order to test all Barth syndrome patients at the meeting plus an equivalent sample of healthy subjects. Recruitment of controls was achieved by word of mouth among friends, families and hotel guests. The synergy between Italian scientists and people of BSF was extremely important for the success of this clinical trial. Drs. Michele Dibattista and Simona Lobasso were able to interact kindly with boys of different ages, put them at their ease, and finally test their smelling ability.

The statistical analysis of data is currently in progress and we plan to soon publish the results of our study. Although relevant olfactory alterations have not been detected, a further in-depth analysis of the patient sample revealed that neutropenic subjects under colony granulocytes stimulating factor are better at discriminating odors.

The smelling test is tricky; scientists have to pay close attention in order to understand if the subject is properly detecting the odors and properly referring his sensation. Patients and controls were very collaborative, despite some technical difficulties. It was moving to see the youngest patients performing the test.



Dr. Simona Lobasso and research
participant, Matthew, during
psychophysical test.
(Photo courtesy of Tiffini Allen 2016)

Finally, I would like to again express all my gratitude to Shelley Bowen and collaborators; without her support we could have not conducted the study. Last but not least, I want to thank Dr. Matthew Toth (BSF Science Director) for his help.



Dr. Michele Dibattista and
research participant, Henry, during
psychophysical test.
(Photo courtesy of Tiffini Allen 2016)

What Are We Going To Do Now?

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

We have many heroes within our community. Ben, at the Barth Syndrome Foundation's (BSF) 2010 conference, said "Give us the treatments and if they don't work we'll try something else." Alas, Ben did not live to be one of our pioneers in clinical trials, but I am sure that if he was writing this article, instead of me, he would be saying more eloquently why these trials are so important. And now we stand at a point we could only dream of a few years ago, trials that could one day help lead to a cure.



Ben
9/2/1994 — 11/17/2010

We all know that Barth syndrome is a bad disease. No one "gets better" with BTHS; in fact, BTHS individuals can and some do die from it. Most Barth syndrome individuals suffer with this disease, and their families do as well. Clinical trials are the significant step in finding a treatment for any disease. Clinical trials are scientific-clinical experiments that are performed on people. That is a scary thought, and it is true that volunteers who participate in clinical trials are taking a risk. They are taking a risk with their health for the benefit of others with their disease, and they do not necessarily benefit themselves. Clinical trial volunteers are heroes.

Our BSF community now faces a turning point in its history. For 2017, clinical trials are planned in the US (Elamipretide with Stealth Biotherapeutics) and in the UK (Bezafibrate with Dr. Colin Steward) to test if either of these two different drugs will benefit BTHS individuals. Volunteers/heroes are needed for these clinical trials. Like all clinical trials, these will not be able to proceed without participants. Our disease may not seem to be as urgent as some other rare diseases with predictable, dire prognoses. Nevertheless, we know BTHS is a debilitating disease that does not get better with time. Doing nothing is not an option. It is difficult to choose to be a hero when it is you or your son at stake, but literally no one other than BTHS guys can do this. Because there are very few BTHS guys in the world (even fewer are eligible due to medical concerns), the pressure is on them. It is "crunch time". It is the bottom of the ninth inning, your side is losing, and people ask, "What are we going to do now?"

This baseball analogy comes up short compared to the clinical trial scenario because historically, 90% of all clinical trials do not end with success—most baseball teams have a better record than that. However, clinical trials have the benefit of a legacy, meaning that each benefits by what was done before, so the first clinical trials will have more unknowns than subsequent ones. Knowing what does not work is often as valuable as finding out what does work. Finding a safe and effective treatment for any disease is a team effort often taking place over many years. It is a job for Team Barth. Will you help? What are you going to do now? What are you going to do for the future? (Photo courtesy of Amanda Clark ²⁰¹⁰)



Brayden (age 7)
(Photo courtesy of Amanda Clark ²⁰¹⁶)



Jacob (age 16)
(Photo courtesy of Amanda Clark ²⁰¹⁶)

Opportunities to Participate in Barth Syndrome Research

A Phase 2 Randomized, Double-Blind, Placebo-Controlled Crossover Trial to Evaluate the Safety, Tolerability, and Efficacy of Subcutaneous Injections of Elamipretide (MTP-131) in Subjects With Genetically Confirmed Barth Syndrome

Details can be found at: <https://www.clinicaltrials.gov/ct2/show/NCT03098797?term=barth&rank=4>

Barth Syndrome Registry

The BRR empowers every person who has Barth syndrome and family members around the world to make a difference in the fight to conquer Barth syndrome. By participating in the BRR and completing your profile survey about your own unique experience with Barth syndrome, you are contributing to a global database about the accessibility of diagnosis, care and treatments, and disease severity of Barth syndrome. The BRR is a centralized resource that is vital to helping researchers learn more about BTHS, accelerating the development of new research and treatments, identifying issues that need research, and improving the care of all those with Barth syndrome.

Do You or a Loved One Have Barth Syndrome?

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 relevant information and contact Dr. Jefferies directly if you decide to help. *(Please see below.)*

The Female Side of Barth Syndrome — Calling All Adult Women!

Dr. Cynthia James and Rebecca McClellan of the Johns Hopkins School of Medicine, Baltimore, Maryland are doing a study concerning how Barth syndrome carrier women navigate the family, reproductive, and psychological implications of being a carrier. Please consider the attached information and contact Dr. James or Rebecca McClellan directly if you decide to help.

As more boys and men are correctly diagnosed with Barth syndrome, mothers, sisters, daughters, and grandmothers face their own challenges. This questionnaire study builds on interviews we did with Barth syndrome carriers to measure the emotional, family, reproductive, and psychological implications of having a relative with Barth syndrome. We are asking ALL ADULT WOMEN to join. We hope study results will help both health care providers and patient organizations provide better care to women in families with X-linked conditions, especially Barth syndrome! *(Please see below.)*

Please visit BSF's website to learn more of these opportunities to participate in Barth syndrome research (<https://www.barthsyndrome.org/about-barth-syndrome/opportunities-to-help-with-barth-syndrome-research>).

Gene Therapy 101: Basic Concepts

By Christina Pacak, PhD, Assistant Professor, Department of Pediatrics, University of Florida, Gainesville, FL

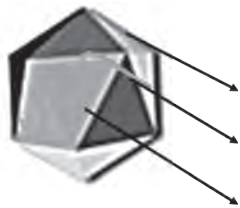


Dr. Christina Pacak
(Photo courtesy of
Dr. Pacak 2016)

[Editor's note: Dr. Pacak received the following research grants from BSF: Correction of mitochondrial dysfunction in Barth syndrome (2014) and Optimization of AAV-mediated gene therapy for Barth syndrome (2016).]

Gene therapy has great potential to treat a variety of disorders caused by single gene mutations. The general goal of all gene therapies is to provide a healthy copy of a mutated gene (*tafazzin* or *TAZ*, in the case of Barth syndrome [BTHS]) to cells that would normally express that gene (in BTHS, heart and skeletal muscle). Since injected genes have a difficult time getting into cells on their own, this process requires approaches where healthy gene copies are packaged into delivery vehicles that help get the healthy gene into targeted cells. For example, certain viruses can easily get into cells and enter the cell nucleus (where genes are housed). These viruses can be used as “carriers” or “vehicles” and allow the gene therapy to “hitch a ride.” Some gene therapy approaches are based on specific gene mutations however, since a variety of mutations exist in different

locations throughout the gene that causes BTHS, we have focused on developing a “one size fits all” gene therapy strategy that we expect will be effective regardless of the underlying *TAZ* mutation.



- Capsid** - determines which cell types are entered
- Promoter** - determines which cell types will express the gene
- Gene** - once inside the nucleus the capsid is removed, DNA becomes double-stranded, healthy RNA is expressed from the gene and translated into the healthy protein!

To achieve the long-term gene expression needed to make the treatment long-lasting or permanent, healthy copies of the necessary gene must be delivered using some sort of vehicle. Amongst the many gene delivery vehicles that exist, adeno-associated virus (AAV) stands out as a highly promising candidate for BTHS for several reasons: (1) it is a non-pathogenic virus with a blunted immune response (AAV exists in nature – we have all been exposed but were unaware of this, as it does not cause sickness); (2) specific serotypes exist in nature that can transduce heart and skeletal muscle (*i.e.*, naturally enter these tissues most affected by BTHS); (3) AAV used in gene therapy cannot insert into the genome, so

there is no worry of disruption of other genes; and (4) AAV-mediated gene delivery provides high level, long-term expression in non-dividing cells (it enters the nucleus and persists as a circular piece of DNA for many years – this can be diluted out in constantly dividing cells such as skin but is not an issue for heart and skeletal muscle where cells do not frequently divide). Importantly, the virus we use cannot replicate on its own, as the viral genes necessary for replication have been completely removed and replaced with a promoter and the therapeutic gene (*TAZ*), thus making it non-pathogenic and safe.

A general consideration for all therapeutics is to establish a minimum effective dose. The objective of this is to expose each patient to the least amount of a therapy needed to achieve a successful treatment response and thus minimize potential side effects, reduce cost per dose, and potentially stretch one batch of a therapy amongst more patients. All of these increase the likelihood of successful translation of a treatment strategy to the clinic. For gene therapy, the way to minimize the effective dose is to strategically consider all of the elements in the delivery vector's design to ultimately achieve strong expression levels limited specifically to those tissues where the gene would normally be expressed.

Thus, to limit *TAZ* gene expression to heart and skeletal muscle, we have carefully chosen the AAV serotype for our delivery vector. Serotypes are slightly different versions of the same virus. The various AAV serotypes each have capsids (outer shells or containers that hold genes to be delivered) with features on their surface that determine their ability to enter specific cell types. We (and others) have previously characterized AAV serotype 9 and found that it does an excellent job of delivering genes to heart and skeletal muscle, so this particular serotype is being developed for BTHS gene therapy. Another way to help limit *TAZ* gene expression to heart and skeletal muscle is through promoter choice. Promoters are DNA sequences that recruit the necessary elements to enable gene expression. The genetic material delivered in gene therapies includes a promoter to drive expression of the healthy gene. Certain promoters are more specific to particular tissues, and, by choosing these specific promoters, we can help restrict expression of the delivered gene to those tissues where it is supposed to be expressed (in BTHS, heart and skeletal muscle). We are currently in the process of identifying the most optimal promoter for BTHS and are pleased that preliminary data have revealed two promising candidates.

Other ways we can optimize gene therapy and minimize the effective dose include a method called codon optimization and the use of double-stranded vectors. Simply, codon optimization is a method to increase the efficiency by which the delivered gene is processed into a functional protein. In addition, since AAV is a single-stranded DNA virus and must become double-stranded following delivery

(Cont'd on page 12)

Gene Therapy 101: Basic Concepts

(Cont'd from page 11)



- (a) Systemic delivery
(throughout the body)
- (b) Coronary delivery
(throughout the heart)
- (c) Direct injection
(localized to a small area)

to cells in order for the delivered gene to be expressed, another vector design consideration is the use of a double-stranded vector designed to enable rapid second strand DNA synthesis and further improve the efficiency of therapeutic gene expression. We have generated BTHS gene therapy vectors that incorporate these modifications and are currently testing their effectivity.

Lastly, we must consider how to deliver the gene therapy to the patient. One option is to directly inject the gene therapy into the heart or skeletal muscle. When the area affected by a disease is contained to a localized region, this may be a good option. However, genes delivered in this manner are only able to reach a limited area surrounding the injection site. For situations such as BTHS where the entire heart and skeletal muscles throughout the body are affected, a broader distribution is needed. Another option (specific to the heart) is delivery through the coronary artery. This method may be considered for BTHS if gene delivery to the heart alone is the goal. A final option, and the one most likely to be used for BTHS, is systemic venous delivery (*i.e.*, through an IV in the arm) which would circulate the therapy throughout the body so that it could target all skeletal muscles and the heart.

With the development of any therapy, it is extremely important to consider patient safety. For gene therapies, there are two main concerns that exist. The first is an immunoresponse to the specific AAV serotype or capsid being used as a result of patients having been previously exposed to that particular AAV. These individuals would essentially be vaccinated against the AAV (even though it never caused sickness) and this could result in the body destroying the therapy before it has a chance to do its job. The second concern is an immunoresponse to

the gene product (*tafazzin*, in the case of BTHS). The problem is that depending upon a patient's particular mutation and whether it results in any protein, the body may identify healthy *tafazzin* as foreign material and raise an immunoresponse. These scenarios are both manageable through preventive administration of immunosuppression (*e.g.*, rituximab) just prior to gene delivery in patients.

Ultimately, the expectation for gene therapies (and our hope!) is to develop treatments in which a single administration provides a beneficial effect for many years. We are steadily making progress on testing the safety and effectiveness of TAZ gene therapy in the BTHS mouse model and in BTHS human cultured cells. We plan to apply for FDA-IND approval and funding for a clinical study within the next year so stay tuned! On behalf of the collaborative team (Dr. Todd Cade, Dr. Barry Byrne, and myself), I would like to close by saying that it has been a pleasure to work with the BSF on this exciting project!



Lindsay Groff, Executive Director, at the table with other nonprofits, representing Barth Syndrome Foundation at the Charity Leader Meeting, part of Better Business Bureau's Wise Giving Alliance. Talking about: cyber security, social media, fundraising, and data sharing. BSF may be a small nonprofit, but we sure are mighty!

Two Retirements from BSF International Scientific and Medical Advisory Board

By Matthew J. Toth, PhD, Science Director, BSF and Katherine R. McCurdy, Founding BSF Board Member

BSF is very fortunate and blessed to have great and good people as part of our International Scientific and Medical Advisory Board, usually abbreviated as SMAB. Dr. Richard Kelley and Dr. Iris Gonzalez are two such outstanding individuals, both of whom have served on our SMAB since the very beginning. Now, each has requested to step back from responsibilities as SMAB members and to officially retire from the board. Both Dr. Kelley and Dr. Gonzalez stepped back, at least partially, from their “day jobs” several years ago but were willing to remain on our SMAB for a bit longer. Now each has decided to make his or her official retirement from the SMAB complete.

Richard I. Kelley, MD, PhD



Dr. Richard Kelley

Dr. Kelley is the physician scientist who, in 1999, offered his lab’s on-line page to three Barth mothers (Shelley, Sue and Anna) as a way for them to reach out to the wider world and to find others affected by the disorder. Then, in 2000, he put together the first small but crucial scientific and medical meeting specifically on Barth syndrome at the Kennedy Krieger Institute and gave families who also had gathered in Baltimore a place to meet for a day. It was a pivotal experience for all who attended, and it was at that meeting that BSF was born. From the beginning, he not only focused his brilliant mind on the complex biochemical issues of Barth syndrome, but he also became a critical clinical resource for many families and their physicians. His insights into topics as disparate as aberrations in amino acid metabolism, management of chronic diarrhea and causes of strong aversion to loud noises, all of which are issues associated with Barth syndrome, are legendary. Dr. Kelley has never been too busy nor too tired to talk with a patient in need anywhere in the world. A number of years ago, he won the “Art of Listening Award” from the Genetic Alliance, and we can understand why, as he is a perceptive and thoughtful listener and a detective when it comes to making sense of complicated symptoms. We asked him to be the first Chairman of our SMAB, and he led this group very ably for many years. We all are indebted to

his attention to detail, his willingness and ability to chart unknown pathways, and his dedication to the boys and men affected by this disorder. *(Photo courtesy of Dr. Richard Kelley)*

Iris L. Gonzalez, PhD



Dr. Iris Gonzalez

Dr. Gonzalez is well-known to many of us, and she is very much a friend. Most know her through her constant presence at BSF conferences where she has explained the genetic complexities of this disease to families in a way that is miraculously understandable. Who can forget her discussion of genetic mutations cleverly presented as a hand-knit sweater with some small mistakes in a stitch or two? Others know Dr. Gonzalez from the meticulous and thorough detail she provides as creator and caretaker of the Barth syndrome genetic database (officially called the Human TAZ Gene Variants Database) for BSF. This is an invaluable resource for many labs, doctors and scientists around the world. Genetics is a subtle but powerful scientific field where more is known each day about how our DNA affects/controls our health and our lives. Dr. Gonzalez provides the scientific-medical world with this solid genetic information about Barth syndrome. In fact, no one on earth knows the *tafazzin* (TAZ) gene as well as she does. Nevertheless, she never forgets that behind each mutation is an individual who suffers, and her insights about the *tafazzin* gene guide our understanding today and will continue to do so in the future. Happily for us, she has agreed to remain on as curator of this database even after her retirement from our SMAB. Those of us who have seen Dr. Gonzalez at work for many years also know

her as a careful, enthusiastic gardener, which figures prominently in the history of genetics.

Drs. Kelley and Gonzalez have each received the Varner award as pioneers in the science and medicine of Barth syndrome. Both were truly instrumental in getting BSF where it is today, and we could not be more grateful for their immense contributions. They are definitely among the host of giants on whose shoulders we now stand. As vital, founding SMAB members, they have greatly contributed to the successes and advances made in understanding and treating this disease. They have helped guide us to our current place of being on the threshold of our first human clinical trials for specific Barth syndrome treatments. We will never forget them, and we hope they will keep in close touch with us, even if not on an official basis. All of us thank them profoundly and wish them the very best. *(Photo courtesy of Amanda Clark)*

Wonderful Volunteer Retires



Les Morris

Les Morris, the grandfather of a young man from Canada who has Barth syndrome, has contributed greatly to the Barth Syndrome Foundation (BSF), quietly and ably by serving on BSF's Publications Team for the last twelve years. Recently, he informed us that he wants to retire from these duties. As a member of the small, international group that works with Lynda Sedefian to edit all the written materials created by BSF he has lent his sharp eyes and his creative talent to making sure that BSF's publications are of excellent quality. In addition, Les served as a board advisor for the Barth Syndrome Foundation of Canada, attending all of their annual planning sessions. Along with his grandson, Adam, Les also volunteered his skills at carpentry, crafting tables, pen cases, etc. that were auctioned off at BSF's international conferences. We are sorry to see him step aside, but we are very grateful for his dedication and commitment to our common cause. Thanks so much for all you have done!

Best wishes for the future, Les, from all in the Publications Team, and we are sure we speak for the Barth syndrome community too. We hope to stay in touch and see you from time to time.

SAVE THE DATE

Barth Syndrome 9th International Scientific, Medical & Family Conference

July 16-21, 2018

Hilton Clearwater Beach Hotel
Clearwater Beach, Florida

Please join us for an exciting two track conference for:

1. **physicians/scientists** unraveling Barth syndrome's mysteries
2. **affected boys/men and their families** (Characteristics: cardiomyopathy, neutropenia, muscle weakness, growth delay, exercise intolerance, 3-methylglutaconic aciduria, cardiolipin abnormalities.)

Learn about the latest scientific developments and clinical insights into Barth syndrome.

Exchange ideas with leading physicians and scientists who are involved in the many aspects of this complex disorder. Network with individuals personally affected and their families.

Help accelerate advances in the understanding and treatment of Barth syndrome and shape the future of Barth syndrome research.

More information available at: barthsyndrome.org

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You Make a Difference

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

"The community is tight-knit, knowledgeable, and inclusive. The Foundation includes not only the patients and their families but also scientists, physicians, and others taking care of Barth patients, all working together - and I mean really together — to cure Barth syndrome. I believe the single "jewel" that epitomizes this Foundation is the biennial international conference they organize, which brings together patients, their families, and scientists from around the world to make headway into this devastating disease." ~ Colin Steward, PhD, FRCP, FRCPCH, Bristol Royal Hospital for Children, England

Barth Syndrome Night at the New York Islanders



Wyatt drops the puck at NY Islanders Game

As you may have read in the last issue, BSF was granted the unique opportunity to have "Barth Syndrome Night" at the NY Islanders game. Wyatt represented us well on the Jumbotron, during the puck drop, and rode the Zamboni! Wyatt could be seen smiling from ear-to-ear throughout the night! Steve McCurdy appeared on Madison Square Garden TV, raising awareness to potentially millions of viewers about Barth syndrome. That night, over \$82,000 was raised to help boys like Wyatt, and all those affected around the world. Huge thanks to the McCurdy family for this connection, to the New York Islanders for hosting us, and to Team Will for volunteering. (Photo courtesy of New York Islanders 2016)

One Couple's Selfless Act on their Big Day



Sarah and Bryan

Sarah and Bryan each have causes that are near and dear to their hearts. Those charities are the Barth Syndrome Foundation for Bryan's son, Abram, and the Michael J. Fox Foundation for Sarah's dad, Wally. Bryan and Sarah love and support each other's efforts because they both know what it feels like to love someone and want them to have a better life. For their wedding day, they asked their friends and family to donate in lieu of traditional gifts. They want to get closer to the greatest GIFT of all, for Abram and Wally to know a cure. **If you or a loved one is getting married and you would like to arrange for donations to BSF, please contact Lindsay Groff at lindsay.groff@barthsyndrome.org.* (Photo courtesy of Bryan 2017)

Bang! Boom! Pow! Devin's League of Superheroes Raises Money for BSF



Devin (age 12)

Devin and his League of Superheroes enjoyed a fun night of hockey with the Flint Firebirds versus the Kitchener Rangers. Ticket proceeds and sponsorships benefitted the Barth Syndrome Foundation. His friends and family, some dressed as superheroes, showed Devin all the amazing support he has in his epic battle against Barth syndrome! Way to go, Devin's League of Superheroes! (Photo courtesy of Nicole 2017)

(Cont'd on page 16)

You Make a Difference

(Cont'd from page 15)

Breaking Barth 2017



Breaking boards for Barth!

To raise money and awareness for BSF, Michael Neece was at it again, breaking boards for Barth! This time with a far larger audience of about 8,000 people. On April 22, 2017 at the University of North Carolina Science Expo, people learned about Barth syndrome and broke a board to symbolize "Breaking Barth." Shelley Bowen, Director of Family Services and Awareness, joined Michael on the stage to raise awareness about Barth syndrome for all attendees on campus. *(Photo courtesy of Michael Neece 2017)*

Happy Heart Week 2017 Raises over \$50,000!



Henry

Sweet Henry celebrated his fifth birthday on May 2, 2017. His loving family chose to honor their adorable boy with a fundraiser for BSF! This Happy Heart Week campaign broke the event's prior fundraising records with over \$50,000 raised in celebration of his milestone birthday. Thank you to all those who gave in Henry's honor, making his birthday all the more exciting. **If you would like to celebrate your birthday with gifts to BSF, please contact Lindsay Groff at lindsay.groff@barthsyndrome.org. (Photo courtesy of Megan 2017)*

A Classic Cycling Event



Jan and Guido

On June 16, 2017 team members Jan, Guido, Ruud, Serge, David, Marco, and Koen participated in the Stykkeproven Trondheim - Oslo Marathon, one of cycling's real classics. Feared and loved by riders for half a century, nothing comes close to completing this race. All money raised went to BSF. *(Photo courtesy of Jan Van Langendonck 2017)*

Financial Advantages To Donating Appreciated Stock

Have you enjoyed a little success in the stock market? Here's something you might not know. If you donate appreciated stock instead of cash, you can take an immediate tax deduction for the full market value of the stock and also avoid the capital-gains tax you'd owe by cashing in the securities. Then, using the cash you might have otherwise donated, you can repurchase the same stock at a higher cost basis for capital-gains purposes. Basically, there are financial advantages to donating appreciated stock instead of cash. For more information please visit BSF's website - Other Ways To Give.

Awareness of Barth Syndrome Continues to Grow

Many Barth syndrome (BTHS) related peer-reviewed journal articles are now being published. To date, a total of **143** 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. Sheeran FL, Pepe S. [Mitochondrial bioenergetics and dysfunction in failing heart](#). *Adv Exp Med Biol*. 2017;982:65-80. doi: 10.1007/978-3-319-55330-6_4.
2. Ikon N, Ryan RO. [Cardiolipin and mitochondrial cristae organization](#). *Biochim Biophys Acta*. 2017 Jun;1859(6):1156-1163. doi:10.1016/j.bbame.2017.03.013. Epub 2017 Mar 20. Review.
3. Menshkyau D. [Emerging technologies for prediction of drug candidate efficacy in the preclinical pipeline](#). *Drug Discov Today*. 2017 May 22. pii: S1359-6446(17)30238-6. doi:10.1016/j.drudis.2017.04.019. [Epub ahead of print]
4. Fajardo VA, Mikhaeil JS, Leveille CF, Saint C, LeBlanc PJ. [Cardiolipin content, linoleic acid composition, and tafazzin expression in response to skeletal muscle overload and unload stimuli](#). *Sci Rep*. 2017 May 17;7(1):2060. doi:10.1038/s41598-017-02089-1. (Open Access)
5. Zhang Y, Bharathi SS, Rardin MJ, Lu J, Maringer KV, Sims-Lucas S, Prochownik EV, Gibson BW, Goetzman ES. [SIRT5 binds to cardiolipin and regulates the electron transport chain](#). *J Biol Chem*. 2017 Apr 30. pii: jbc.M117.785022. doi: 10.1074/jbc.M117.785022. (Open Access)
6. Finsterer J, Stollberger C. [Acquired noncompaction in Barth syndrome due to the TAZ mutation c.481_482ins20](#). *J Pediatr*. 2017 Mar 15. pii: S0022-3476(17)30332-3. doi: 10.1016/j.jpeds.2017.02.066. [Epub ahead of print]
7. Bakšienė M, Benušienė E, Morkunienė A, Ambrozaitė L, Utkus A, Kucinskas V. [A novel intronic splice site tafazzin gene mutation detected prenatally in a family with Barth syndrome](#). *Balkan J Med Genet*. 2017 Mar 8;19(2):95-100. doi: 10.1515/bjmg-2016-0043.
8. Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. [The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome](#). *Orphanet J Rare Dis*. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. (OpenAccess)
9. Lloyd DF, Vara R, Mathur S. [The cardiac manifestations of inherited metabolic diseases in children](#). *Pediatr Int*. 2017 Mar 4. doi: 10.1111/ped.13272. [Epub ahead of print] Review.
10. Schlame M, Xu Y, Ren M. [The basis for acyl specificity in the tafazzin reaction](#). *J Biol Chem*. jbc.M116.769182. doi:10.1074/jbc.M116.769182. (OpenAccess)
11. Bashir A, Bohnert KL, Reeds DN, Peterson LR, Bittel AJ, de Las Fuentes L, Pacak CA, Byrne BJ, Cade WT. [Impaired cardiac and skeletal muscle bioenergetics in children, adolescents, and young adults with Barth syndrome](#). *Physiol Rep*. 2017 Feb;5(3). pii: e13130. doi: 10.14814/phy2.13130.Δ
12. de Taffin de Tilques M, Tribouillard-Tanvier D, Tétaud E, Testet E, di Rago JP, Lasserre JP. [Overexpression of mitochondrial oxodicarboxylate carrier \(ODC1\) preserves oxidative phosphorylation in a yeast model of the Barth syndrome](#). *Dis Model Mech*. 2017 Feb 10. pii: dmm.027540. doi: 10.1242/dmm.027540. [Epub ahead of print]
13. Wang J, Guo Y, Huang M, Zhang Z, Zhu J, Liu T, Shi L, Li F, Huang H, Fu L. [Identification of TAZ mutations in pediatric patients with cardiomyopathy by targeted next-generation sequencing in a Chinese cohort](#). *Orphanet J Rare Dis*. 2017 Feb 10;12(1):26. doi: 10.1186/s13023-016-0562-4.
14. Sathappa M, Mitchell W, Coscia A, Boyd K, May E, Szeto HH, Alder NN. [Investigation of the interactions of the SS-31 peptides with cardiolipin variants: A potential therapeutic for Barth syndrome](#). *Biophysical Journal*, Volume 112, Issue 3, Supplement 1, 3 February 2017, Pages 438a.
15. Dudek J, Maack C. [Barth syndrome cardiomyopathy](#). *Cardiovasc Res*. 2017 Feb 2. doi: 10.1093/cvr/cvx014. [Epub ahead of print]
16. Kagan VE, Bayir H, Tyurina YY, Bolevich SB, Maguire JJ, Fadeel B, Balasubramanian K. [Elimination of the unnecessary: Intra- and extracellular signaling by anionic phospholipids](#). *Biochem Biophys Res Commun*. 2017 Jan 15;482(3):482-490. doi: 10.1016/j.bbrc.2016.11.005. Epub 2017 Feb 3.*
17. Jacob ML, Johnco C, Dane BF, Collier A, Storch EA. [Psychosocial functioning in Barth syndrome: Assessment of individual and parental adjustment](#). *Children's Health Care*, 46:1, 66-92 (2017).
18. Borna NN, Kishita Y, Ishikawa K, Nakada K, Hayashi JI, Tokuzawa Y, Kohda M, Nyuzuki H, Yamashita-Sugahara Y, Nasu T, Takeda A, Murayama K, Ohtake A, Okazaki Y. [A novel mutation in TAZ causes mitochondrial respiratory chain disorder without cardiomyopathy](#). *J Hum Genet*. 2017 Jan 26. doi: 10.1038/jhg.2016.165. [Epub ahead of print]
19. Schlame M, Greenberg ML. [Biosynthesis, remodeling and turnover of mitochondrial cardiolipin](#). *Biochim Biophys Acta*. 2017 Jan;1862(1):3-7. doi:10.1016/j.bbali.2016.08.010. Review.
20. Shen Z, Li Y, Gasparski AN, Abeliovich H, Greenberg ML. [Cardiolipin regulates mitophagy through the PKC pathway](#). *J Biol Chem*. 2017 Jan 5. pii: jbc.M116.753574. doi:10.1074/jbc.M116.753574. [Epub ahead of print] (Open Access)
21. Woiewodski L, Ezon D, Cooper J, Feingold B. [Barth syndrome with late-onset cardiomyopathy: A missed opportunity for diagnosis](#). *J Pediatr*. 2017 Jan 18. pii: S0022-3476(16)31565-7. doi: 10.1016/j.jpeds.2016.12.070. [Epub ahead of print]
22. Mejia EM, Zinko JC, Hauff KD, Xu FY, Ravandi A, Hatch GM. [Glucose uptake and triacylglycerol synthesis are increased in Barth syndrome lymphoblasts](#). *Lipids*. 2017 Jan 17. doi: 10.1007/s11745-017-4232-7. [Epub ahead of print]*
23. Ikon N, Ryan RO. [Barth syndrome: Connecting cardiolipin to cardiomyopathy](#). *Lipids*. 2017 Jan 9. doi: 10.1007/s11745-016-4229-7. [Epub ahead of print]
24. Tyurina YY, Lou W, Qu F, Tyurin VA, Mohammadyani D, Liu J, Hüttemann M, Frasso MA, Wipf P, Bayir H, Greenberg ML, Kagan VE. [Lipidomics characterization of biosynthetic and remodeling pathways of cardiolipins in genetically and nutritionally manipulated yeast cells](#). *ACS Chem Biol*. 2016 Dec 16. [Epub ahead of print]*
25. Dong X, Fan P, Tian T, Yang Y, Xiao Y, Yang K, Liu Y, Zhou X. [Recent advancements in the molecular genetics of left ventricular noncompaction cardiomyopathy](#). *Clin Chim Acta*. 2016 Dec 15;465:40-44. doi:10.1016/j.cca.2016.12.013. [Epub ahead of print] Review.
26. Raja V, Joshi AS, Li G, Maddipati KR, Greenberg ML. [Loss of cardiolipin leads to perturbation of acetyl-CoA synthesis](#). *J Biol Chem*. 2016 Dec 9. pii: jbc.M116.753624. [Epub ahead of print]*

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Barth Syndrome Trust — Update from Chair

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



Nick & Michaela
(Photo courtesy of
Amanda Clark 2016)

It was a great pleasure to represent our International Barth syndrome community as a guest speaker at the recent Genetic Disorders UK Leadership Symposium in London. This is always a wonderful meeting as the leaders of all the genetic disorder charities, other patient advocates, and various experts in the field gather to share experience and to learn from each other. The event was hosted by Genetic Disorders UK in partnership with Global Genes. With a carefully thought out agenda, the day provided a wealth of valuable information as well as a host of inspirational stories from like-minded people. My talk focused on the importance of people in our battle against our ultra-rare condition. (<http://www.geneticdisordersuk.org/leadershipsymposium/2017leadershipsymposium/presentations#michaeladamin>)

Barth Syndrome Trust — Looking Ahead

The Barth syndrome community is incredibly fortunate to have such a dedicated and talented group of people all around the world who devote their time and their money to creating a better future for those with Barth syndrome. Busy people, stressed people, tired people who still respond when asked if they can help.

As the Barth Syndrome Trust plans for the next few years, I have a few key questions for you.

- What can we do for you?
- What can we do better?
- What should we be doing differently?

We would love to hear your ideas about how we can improve — drop us a comment on our Facebook Family Page or via email on info@barthsyndrome.org.uk.

Most of our families are already doing all the things below, but if you are new to Barth syndrome or wondering how best you can help us to make positive changes, then it would be wonderful if you would help us out by doing some or all of the things below. Conversely, if you run into problems or have questions, please let us know what barriers you face so that we can better understand and help.

- Join the registry (www.barthsyndromeregistry.org). We need everyone's data to map out the true picture of Barth syndrome.
- Sign up to participate in clinical trials and research — we are so few in number. Without you we cannot succeed.
- Attend clinics, outreaches and meetings.
- Think about making a donation, doing a fundraiser, or volunteering some of your time.
- Share your story and your experience.

UK Family Barth Fun Weekend: 9th-11th June 2017

In the past, we always arranged a Fun Day at the end of each clinic. Now that the clinics are run more often, but with fewer people attending each time, there are very few opportunities for Barth families to meet up. This year we are hosting one big Family Outreach for all Barth families.



The national Barth weekend in the New Forest (www.avontyrrell.org.uk) will be filled with fun, challenging but safe activities like abseiling, zip wire, swimming and kayaking. Special adaptive equipment means that the activities will be accessible to all. So far, we have around 75 people attending, so it will be a great chance to relax in a beautiful setting and have fun together.

Many thanks to Jeans for Genes for providing us with a grant towards the cost of this weekend. We can't wait to see you all! Look out for reports and photos of the weekend in the next newsletter.

News from the Bristol Barth Syndrome Service

By Brenda Harding, on behalf of the Barth Syndrome Service Team

A fond farewell to Debbie Riddiford...

A fond farewell to the Barth Syndrome Service's wonderful nurse, Debbie Riddiford, CNS, who retired at the end of March 2017. We are so grateful for her massive contribution to the successful running of the Barth Syndrome Service over the past seven years. Happily, Debbie will be coming back for the next few clinics to help us out in this transition period.



Prof. Colin Steward & Debbie Riddiford



Hayley Smith, CNS

Hello Hayley...

The role of Clinical Nurse Specialist (CNS) is being filled in the meantime by Hayley Smith who worked with Debbie to ensure a smooth handover. Hayley is a very experienced CNS having worked for many years for the Home Ventilation team at Bristol Royal Hospital for Children. We will introduce Hayley to you all when you attend future clinics, but if you need to contact her she is available through PKB or on 07795 507 294.



Lucy Buckley

Goodbye Lucy, hello Livvy...

March also saw us saying goodbye to our wonderful physiotherapist, Lucy Buckley, as she started her maternity leave. We congratulate Lucy on the birth of her beautiful baby daughter, Nia, on 16th April. We can't wait to meet her. Lucy's post is being covered by a new physiotherapist, Livvy Berry, who joined us on 1st May. Livvy attended the clinic on 5th May, so some of you will have met her then. She is available through PKB or on 0117 342 8525.



Livvy Berry



Dani Goodman

And hello again to Dani Goodman...

We are also pleased to tell you that Dani Goodman will be providing occupational therapy at the next few clinics and we are expecting that she will be able to return fully to the service in the Autumn.

This is a period of transition for the Barth Syndrome Service, but please be assured that the team is here for all of you. If you have any queries, please contact us through PKB or speak to Brenda Harding, our Administrator, who will happy to pass on any messages to the relevant team member.



Brenda Harding

As many of you already know, Prof. Colin Steward is due to retire towards the end of 2017, and we'll explain more about the Consultants who will be taking over for him in the coming months. On the research side, he intends to continue running the CARDIOMAN (Bezafibrate) drug trial when it starts next winter, so luckily we won't be seeing the last of him for a good while yet! (Photos courtesy of BST 2017)

Nicholas's Kitchen

By Miriam Pape, Corporate Responsibility Coordinator, Treasury Wine Estates, Australia



Pam Holmes

Pam Holmes, an employee from Coldstream Hills Winery, has set up a registered catering business called Nicholas's Kitchen, where she prepares meals and an assortment of pickles and chutneys for the permanent and casual employees at Coldstream Hills. Through this initiative, Pam has raised AUD\$800.00 for the Barth Syndrome Trust UK & Europe. In February 2017, Pam accessed Treasury Wine Estate's 1124 Gift Program and was able to match the money raised taking the total donation amount to AUD\$1,600.00.

The Barth Syndrome Trust has set the goal of searching for a cure to Barth syndrome, a genetic disorder affecting boys/men that causes heart failure and a risk of sudden cardiac arrest. The Trust works to get more children correctly diagnosed, supporting families by providing information, and funding research into treatments and the search for a cure.

Michaela Damin, the Barth Syndrome Trust Chairperson, penned a letter on behalf of the Trust to thank Pam and TWE for the donation:

"We have been incredibly fortunate to have the support of fundraisers like Pam Holmes who work so hard to raise money to support our programs and, having her donation matched by the company is a wonderful way to maximize the impact of her fundraising."

TWE's 1124 Gift Program offers employees the opportunity to have their fundraising efforts matched up to the value of AUS\$1124.00. (Photo courtesy of Treasury Wine Estates 2016)

Inspired by the 2017 Genetic Disorders UK Leadership Symposium

By Brandon Tang, Westminster School, London

Our school has always wholeheartedly supported its pupils' efforts to raise funds for charities. But although we lend our support to organizations that advocate well-known, global causes like gender equality and the prevention of HIV, we are not as well-informed of the myriad rare genetic conditions that collectively affect so many people around the world, but whose afflicted and their families do not receive as much attention and support. That was the feeling we got as we witnessed chairpersons, founders, and directors of numerous charitable organizations, both international and local, enthuse about their work and their aspirations at the 2017 Genetic Disorders UK Leadership Symposium. It was very invigorating for us to hear about all the hard work people have put into projects that allow patients with rare disorders and their families to navigate their daily lives more easily. It is also incredibly admirable that the charities remain so dedicated to making the most out of their strained resources.

Of these charities, it was Barth Syndrome Trust (BST) that impressed us the most, having started only 16 years ago, and currently with only 26 known affected individuals in the UK. BST and its partners are now on the brink of the first two clinical trials for Barth syndrome. How the patient families have reached out to each other and help form an intercontinental but close-knit community was nothing short of amazing for us. In fact, as subsequent speakers mentioned initiatives for fundraising and raising awareness, we had already decided that we should try to make a difference to this worthwhile cause. Although our school had only just concluded a huge fundraising campaign for another charity, we nonetheless decided to organise a casual dress day dedicated to rare genetic conditions like Barth syndrome. We talked about Barth syndrome and the BST in a school assembly and held a charity book sale as well.

Even though we all have yet to meet someone affected by Barth syndrome, this small fundraising initiative has really allowed us and our schoolmates to learn about this disease and the brave people who have all come together to stand up to it, and we all wish you success in finding a successful treatment for Barth syndrome soon.

Thank you, Westminster School, for the amazing sum of £1236.25.

Barth Syndrome Foundation of Canada President's Report

By Susan Hone, President, Barth Syndrome Foundation of Canada

Like many others in their articles in this issue, I would like to encourage all of our Barth family to participate in the clinical trials upcoming this year and in the near future. I am amazed and excited that we have hit these major milestones so quickly. The scientists and doctors have done their part, and now it is our turn. We asked for possible treatments and/or a cure, and they have delivered. The next steps are up to us, we must volunteer for the clinical trials.

To the doctors and scientists who have worked so hard to make these trials a reality, I thank you with all my heart. You are amazing and I feel honoured to have met so many of you.

To all the families who have done numerous fundraisers over the years to fund the research, I thank you as well.

Giving Tuesday was once again successful in raising over \$12,000. We were very fortunate to have two matching donations made. Our annual spring appeal is in the works, and we have once again been offered a matching donation to entice donors.

Barth Syndrome Foundation of Canada (BSFCa) was again able to contribute \$25,000.00 towards the annual grant process sponsored by the Barth Syndrome Foundation. This year, we are sponsoring Christina Pacak, PhD, Assistant Professor University of Florida, Gainesville, Florida "Optimization of AAV-mediated gene therapy for Barth syndrome." (For more information, please see pgs. 1/4-5)

The chart below shows BSFCa's financial information for the year ended December 2016.

| | |
|--|-------------|
| Revenue | \$ 47,168 |
| Operating Expenses | \$ 12,423 |
| BSF Conference Involvement (this includes physician flights and accommodations) | \$ 8,649 |
| Research Grant Funding | \$ 38,304 |
| Net Revenue | (\$12,208)* |

*Note: As we had a healthy bank balance of \$110,000 at the end of the 2015 fiscal year, the Board made the conscious decision to have a negative net revenue in order to fund the research grant and contribute to the BSF conference in 2016, leaving us with a still strong bank balance of just under \$100,000.

I look forward to seeing what accomplishments our Barth family makes in 2017.

Barth Syndrome Foundation of Canada Getting the Word Out on Research Grants

By Susan Hone, President, Barth Syndrome Foundation of Canada



Dr. Robin Duncan
(Photo courtesy of
Dr. Duncan 2016)

I recently asked one of our grant recipients, Dr. Robin Duncan, Assistant Professor, Physiology & Nutrition, University of Waterloo, about how she knew about the Barth Syndrome Foundation (BSF) Research Grant Program and how to get the word even farther out into the research community. While the answers are from a Canadian researcher (and edited for length), I am sure they would apply to researchers worldwide.

How did you hear about BSF's Research Grant Program? I'm not entirely sure where I first heard about BSF's Research Grant Program – likely it was from several sources around the same time. I know I heard about them from a colleague who was a recipient of a BSF grant. Researchers are very excited about foundation grants, because they are so targeted to specific research, whereas the general competitions are untargeted and in the present day, funding is very tight (and success is very low, even for well-established,

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Barth Syndrome Foundation of Canada

Getting the Word Out on Research Grants

(Cont'd from page 22)

world-renowned researchers). As a new researcher, I recall at least two, if not three, other researchers in the phospholipids field telling me about BSF as soon as they realized I was working on phospholipids and cardiolipin.

In the same way, I have had colleagues offer general advice that whenever I work on any biochemistry that underlies a disease, I should check to see whether that disease has a foundation.

Finally, we get information from the university's central Office of Research every month, about foundations and granting agencies that have active competitions. This is probably the single best way to reach researchers working in fields related to Barth syndrome.

What prompted you to apply for the grant? Need is always the driver. I had accumulated a substantial body of data showing that we had an enzyme that was somehow increasing cardiolipin. Ten years ago, I am convinced my data would have been sufficient for a major government grant. Today, though, all research is underfunded, and you essentially have to have partial answers to all of your questions to get funding. As a new researcher, I was afraid that I would not be able to continue this work. This grant opportunity has allowed me to continue the research, with the goal of achieving major funding. The moment we started seeing effects on cardiolipin, I thought "Okay – I know there is a foundation out there – something to do with *tafazzin* – we need to find them." I waited until we had enough data to make a compelling case, and also until we needed to secure resources or would have to drop it!

This paradigm very much works in your favour right now – there is enormous need, and not enough resources from traditional sources. Researchers will certainly find you! Anyone with a promising lead will be looking for ways to keep the work alive. Anyone who has found the resources (*e.g.*, major government grants) will talk to you (and the world) about it, but won't apply for grants.

Where do you think we should advertise BSF's Grant Program in Canada? I would approach research universities across Canada. They all have an Office of Research that will disseminate your opportunities to the entire campus. I wouldn't spend money on advertising. For one thing, researchers are good at research. We will uncover opportunities by searching ourselves. We don't tend to be reached by traditional media. Anyone who is established and working in cardiolipin already knows about BSF. Anyone who is new to cardiolipin research would talk to established cardiolipin researchers, who would tell them about BSF.

BSF is in a particularly fortunate place, in that although Barth syndrome is exceptionally rare, changes in mitochondrial cardiolipin are incredibly common in the majority of chronic

diseases, including cancer, Alzheimer's disease, and even normal aging. There is, therefore, a huge amount of scientific interest in Barth syndrome as a model for understanding cardiolipin metabolism and mitochondrial metabolism in health and disease. Work on cardiolipin, *tafazzin*, ALCAT1, etc. – it all has implications for virtually everyone on the planet. As a result, everyone receiving a catalyst grant from BSF stands a good chance of getting major governmental funding because of the implications for health of the entire population.

Do you know of anyone presenting at conferences, meetings, etc. that would be willing to speak about Barth syndrome? Do you know of any yearly conferences we should be targeting? Barth syndrome researchers, and anyone working on cardiolipin, will present at conferences and meetings and will definitely want to talk about this disease. It may be worth asking researchers if they would specifically talk about personal aspects of the disease and its severity, to try to raise awareness. I think that both Ryan Bradley and I found that attending the Barth Syndrome International Scientific, Medical & Family Conference in Florida had a major impact on our emotional commitment to involvement with Barth syndrome research. Ryan has been wearing his blue wristband ever since! We will be going on a Barth syndrome-inspired tour of the universe just as soon as we submit our work for publication.

In terms of targeting, there are a lot of different conferences, which makes it difficult to say. There are huge annual conferences like experimental biology (EB), but it is difficult to get a message across in these, and there tends to be a lot of smaller sessions running concurrently (it would be hit-and-miss in terms of advertising). There are also smaller conferences that may allow greater targeting. There are so many though, that I'm not sure that advertising with them would be effective either. The best approach seems to be having researchers talk to each other, which we already do. And since we are always looking for the means to pursue research, we tend to share this information liberally.

Any other suggestions on how to spread the word about the Research Grant Program or Barth syndrome in general would be great. I would perhaps encourage BSF supported researchers to consider speaking about their personal experiences from attending the BSF conference. That sort of personal connection helps to engage the audience (even a scientific one) and generally helps the audience to pay attention and remember what they heard!

William in the News

Six year old William, a Canadian affected with Barth syndrome, was recently in his local news.

To read about William (in French) follow this link: <http://www.lerefletdulac.com/actualites/2017/4/18/william-bernard--l-enfant-qui-fait-rayonner-l-estrie.html>.

There is another article at this link as well: <http://www.lapresse.ca/la-tribune/actualites/sherbrooke/201704/11/01-5087510-enfant-soleil-des-fonds-pour-aider-des-enfants-malades-comme-william.php>.

Good job William! (Photo courtesy of Jennefer 2017)



William holds on to his dad

Remembering Andrew

By Susan Hone, President, Barth Syndrome Foundation of Canada

Andrew Hope, son to Chris and Michael, twin brother to Robert, and brother to David and the late James, passed away unexpectedly on January 2, 2017. Although Andrew did not have Barth syndrome, he had his own health challenges, one being cerebral palsy.

Andrew always greeted me with a huge smile anytime I saw him. He was also very wary of me as he knew when I showed up it usually meant I was taking his mother away (to Barth syndrome planning meetings), and he was not impressed by that. When we returned after our meeting, I would get the cold shoulder for the first day. Andrew loved when my son, Jared, would come visit. They would listen to books, watch movies, and communicate with each other. They were great friends.

To all the Hopes, I am so sorry for your loss. Andrew will be in my heart forever.

End of An Era

By Susan Hone, President, Barth Syndrome Foundation of Canada

Once in a lifetime, you meet someone who is as close to the perfect friend, volunteer, and mentor as you will ever know. I met that person the day I met Lois Galbraith.

Lois and I met at a Barth Syndrome Foundation (BSF) Volunteer Workshop in November of 2004. I came away thinking "WOW, what an amazing woman," and I was right. Lois has been there at a moment's notice for Barth Syndrome Foundation of Canada (BSFCa) and BSF for over ten years. She has been involved in almost every aspect of our foundation — secretary, family services, and volunteer recruitment to name a few. She has been the familiar face at the welcome desk for the Barth Syndrome conferences. There are just too many examples of how Lois has become the person to turn to in need.

Lois recently informed BSFCa that she is no longer able to volunteer as she previously has. Although I will miss calling her to ask for "one more favour," I wish her happiness and health in the next adventures she and Les undertake. On behalf of BSFCa, thank you, Lois, for everything you have done, your never-ending support and your love of our Barth family. (Top photo courtesy of Amanda Clark 2016; Bottom photo courtesy of Tiffini Allen 2016)



Lois Galbraith and Adam (age 27)



Felicia Ganote, Lois Galbraith and Sharon Olson at BSF's 2016 conference

Association Barth France

Why I Need TeamBarth to Come Together

By Florence Mannes, Chair, Association Barth France



Raphaël (age 7) & Florence
(Photo courtesy of Amanda Clark 2016)

Every six months, I have the same ritual: the kids are asleep, the house is quiet, so I can dig into my "Barth documentation" to find the subject of the article I will write for the BSF Journal.

Usually, I just give some updates on the events Barth France had organized during the past six months...The Charity Dinner, the Poker Tournament, the Golf Tournament, the multiple races... one issue after the other. I always have the feeling that I'm roughly writing the same stories.

That's why this time I decided to first have a look at every single article I had written for the BSF Journal since the very beginning of Barth France in 2010. I was really surprised to see where we are now and where we came from...such a long way in only six years!

From a 4-member association to 1,200+ donors, from a 6-runner team to a team of more than 80 marathoner runners, from a single French Barth Family to a 22 Barth cases cohort, from ideas of treatments to actual clinical trials...

In the Fall/Winter issue of 2011, I had set three goals for the next three years: (1) a better diagnosis of Barth syndrome (BTSH) in France; (2) an increased knowledge of BTSH by the doctors; and (3) an expansion of French applications for BSF research grants (French scientists who would apply for BSF research grants)... Well, it took us a bit longer, but here we are! These goals have been achieved, and, not only did a French scientist apply for a BSF research grant, but she received it this year!

We, along with the Barth Syndrome Foundation, have recently made tremendous progress; we have never been so close to finding treatments, never been so close to insuring our boys a better life...

However, despite all that has been done, I'm fearful...fearful that we might never manage to reach our ultimate goal, which is to find a cure for Barth syndrome.

I fear that everything we have done so far would be useless...What if there are not enough boys or men who volunteer for clinical trials? What if all the great ideas scientists come up with remain buried in labs because we lack volunteers?

We, Barth families, are a tiny community, and we deeply rely on each other; our team consists of approximately 200 affected boys and men around the world; each trial has its specificities, and only some of us are eligible for each one. In order to move forward, we need everyone who is eligible to participate...we are a team...

We, Barth France, as a French association, as a family, can organize events, raise money, run to raise awareness...and that's what we do...

But there is one thing we **CANNOT** do on our own: we **CANNOT** participate alone in the clinical trials; everyone has to be involved, we need everyone.

Recent Events

In the meantime, we are still doing our best to raise funds and awareness...and we will do so until a cure is found.

Over the last months, we organized the 5th Edition of the Barth France Poker Tournament, the 3rd Edition of our Black Truffle Charity Dinner, and we raised as a whole in 2016 around \$100,000 USD, which allows us to fund a BSF research grant, allocated to a French scientist. (For more information, please see pages 1/4-5.)

We continue to have many runners participating in several races, and we even had a 4-minute TV show about Barth syndrome on a national channel, again, to raise awareness.

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Association Barth France

Why I Need TeamBarth to Come Together

(Cont'd from page 25)

Though we love what we do, I really hope that all our efforts will not be in vain due to a lack of clinical trial participants.

So, as I did in 2011, let's set some objectives for Barth France for the coming years...

- More French families attending the BSF conference in 2018
- More French scientists working on Barth syndrome in 2020
- Barth syndrome clinical trials in France and in Europe in 2022

Let's come together!



Just before the arrival of the 200 invitees to Barth France's 3rd Edition of the Black Truffle Charity Dinner



Philippe congratulates Chef Hervé for cooking for the 200 people attending Barth France's 3rd Edition of the Black Truffle Charity Dinner



5th Edition of the Barth France Poker Tournament



Vincent Corlay, Pietro Anselmi and Didier Brignand — start of the London Marathon

(Photos courtesy of Barth France 2017)

Barth Italia

Two Happy Children Despite Tough Times

By Elina & Catalin, Parents of Affected Individuals (7 months & 4 years of age), Italy



Matei (age 4)
(Photo courtesy of Elina 2017)

We have two boys with Barth syndrome. Matei was born on the 27/10/2012. At the age of 14 months, he was diagnosed with dilated cardiomyopathy after experiencing a common cold. Up to that age, he never experienced health issues and developed normally. But at just over a year old, he was placed in the hospital for a long period of time. The doctors couldn't find the cause of the sickness; they presumed it was an untreated myocardial infection. Matei's heart failure was serious, and there was talk about a heart transplant. Fortunately, his heart improved as the result of medication.



Teodor (age 7 months)
(Photo courtesy of Elina 2017)

Our "Barth" story really began, though, in March 2016 when Matei had a cardiogenic shock. For many days, he was in intensive care on artificial ventilation, sedated and full of tubes. The doctors were not able to reduce his temperature; he suffered 39 °C (equivalent to 102.2 °F) for 14 consecutive nights. I paced through unbearable moments, as I watched my child being taken away from me. All I could do was suffer and ask the Lord to save him. He was placed on the heart transplant emergency list. Pediatricians observed both neutropenia and dilated cardiomyopathy and suspected that Barth syndrome was present. They decided to go forward with G-CSF, and, in a short time, his neutrophils increased and regulated his temperature back to normal. Genetic tests confirmed the diagnosis of Barth syndrome. Receiving the diagnosis was truly a painful moment and difficult to accept, but we finally had a full diagnosis and more perspective on the various components of the illness. Matei's heart began to function properly, and he was taken off the transplant waiting list.

During all this time, inside my womb grew a beautiful baby, Teodor. His presence gave me strength and most importantly, hope. I prayed that the genetic mutation would not be passed onto him, but it was. He was born on 23/09/2016, and he developed health problems immediately after birth, coming into this world with severe hypoglycemia, heart failure, and difficulty breastfeeding. He was transferred to neonatology unit (often called the NICU), and, after the first eight days of his life, he reached the stage of septicemia. This affected all his internal organs, but fortunately did not create permanent problems. For a long period of time, he was in intensive care, just like his brother. He was forced to fight for his own life. Everything seemed to be a nightmare that we prayed would end so we could go home. When I left the hospital with Teodor in my arms, I was happy and emotional about the brothers meeting. The moment he met Matei was like magic.

After many hours spent searching the internet, I came across the Muller family, and their son, Pietro, and the existence of the Barth Italia Association. I found a telephone number and rang. A boy with a warm voice picked up the phone. I immediately assumed it was Pietro. After that, I managed to get in contact with his mother, Paola, an amazing woman. We immediately bonded with each other. We spoke a lot. Our stories were very similar. I had finally found someone who understood the pain I had been through. She provided me with information about this illness and how to manage it. Finally we met, and being with Pietro and his family made us very happy. Pietro is a wonderful teenager, and his parents are amazing people. Through this association, I met other families from Italy and around the world. I've come to know the volunteers, activities, and the association's projects.

For Christmas, the association organised a fair to raise funds. Matei loved meeting Santa, he even made a drawing and wrote a letter for him.

Also, the charity dinner was a special moment. I was surprised by how many people supported us. Then, with the Muller family, we also shared a special event, Teodor's baptism.

Matei and Teodor are two amazing young boys. Despite all the tough times, they are two happy children. Thanks to the doctors, family, friends, the association, and everyone who supported us.



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Barth syndrome (BTHS; OMIM #302060) (ICD-10: E78.71)

A rare, life-threatening genetic disorder primarily affecting males around the world. It is caused by a mutation in the *tafazzin* gene (*TAZ*, also called G4.5), resulting in an inborn error of phospholipid 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** (*can be chronic, intermittent, cyclic, or not present*)
- **Low muscle mass and muscle weakness**
- **Growth delay** (*short stature in the early years, followed by accelerated growth in mid- to late puberty*)
- **Exercise intolerance** due to early fatigue
- **Feeding problems** (*e.g., difficulty sucking, swallowing, or chewing; aversion to some food textures; selective or picky eating; frequent vomiting*)
- **Cardiolipin abnormalities**
- **3-methylglutaconic aciduria** (*variable but typically a 5- to 20-fold increase*)



Bryn (age 5)



Noah (age 15)
(Photos courtesy of Amanda Clark 2016)