Barth Syndrome Foundation®
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Barth Syndrome Journal

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

2016 Science and Medicine Sessions A Glimpse of the Final Frontier?

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

The 2016 Barth Syndrome International Scientific, Medical & Family Conference was remarkable. The motto for the conference, "Team Barth," brought into focus what this new frontier of clinical trials/studies will mean for Barth syndrome individuals, their families, and the researchers and clinicians who attended. Like previous conferences, the 2016 meeting provided information for new and for experienced families, collected new scientific/clinical information, and offered valuable training/lifestyle tips. The Scientific and Medical sessions (SciMed) showcased the incredible scientific progress in understanding Barth syndrome and outlined the therapeutic ideas and therapies that have germinated with BSF's support over the years. The BSF community is now at the stage where Barth syndrome individuals need to volunteer for clinical studies and clinical trials to reach our common goal. It will certainly take a "team effort" to reach this "final frontier" that we hoped would come one day.

Over 85 scientists and physicians attended the SciMed sessions, the largest such gathering at these biennial conferences. As in previous years, many of the presentations were recorded on video and are available for viewing on the BSF website (https://www.barthsyndrome.org/science--medicine/-scientific--medical-presentations/july-21-22,-2016---scientific--medical-sessions). These videos are a great refresher for the participants and a valuable resource for those who were unable to attend. One can also trace the advancements in our understanding of Barth syndrome over the years and how they are being translated into possible therapies by viewing the extensive collection of these conference videos. *(Cont'd on page 4)*

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Our Brand New Threshold of Hope — Clinical Trials

By Sue Wilkins, RN, Former Board Member, Barth Syndrome Foundation

As a community, we have come so far in the last 16 years. From our first gathering in 2000, when a group of parents from all over the world bravely came together and then made the decision to create the Barth Syndrome Foundation (BSF), to our hope-filled BSF conference this last July. From the very beginning, our goal has been finding treatments and a cure for this disorder. ***And now we are actually approaching that time and are the closest we have ever been to finding treatments for Barth syndrome***. Our Barth community is literally standing on a new threshold of hope and our future. It's a threshold of something many of us have dreamed about for decades — in my case, 34 years. For the first decade of my son's life, this disorder didn't have a name, much less the hope for any potential treatments. And now we are **here**. Let's talk about it!

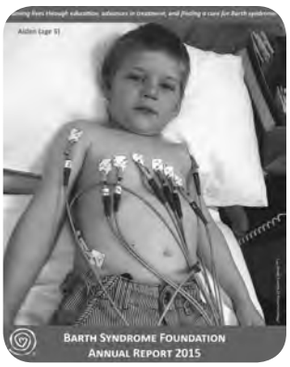
At the BSF Conference in July, Kate McCurdy (BSF SMAB *Emerita*) and Marc Sernel (BSF Board Chair) gave a presentation about a number of potential therapies for Barth syndrome that are in the works, as well as upcoming clinical trials for these therapies. They informed us that two of these therapies could be moving toward clinical trials within the next year. Friends, THIS IS A BIG MOMENT for our entire community. *(Cont'd on page 7)*

The Epic Bravery that Inspires Barth Syndrome Progress

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

I love meeting our donors. On October 24th, about 75 people gathered to hear about our work at the Barth Syndrome Foundation and to learn about regenerative medicine and its application to Barth syndrome.

That night we heard fascinating talks by Dr. Doug Melton and Dr. Bill Pu from the Harvard Stem Cell Institute. As our guests made their way toward the door afterwards, we distributed some materials, including our 2015 annual report.



Aiden (age 5)

A longtime BSF donor whom I hadn't yet met in person, picked up the report and told me how it moved her when she received it in the mail earlier this year. She explained that the photo of adorable little Aiden, age 5, on the cover stirred something inside of her. She picked it up, pointed to the photo and said, "Look at his eyes…see how he's putting on that brave face. I got emotional, thinking about how hard it is for him, yet how determined he is." A brave face indeed.

This symposium fell on the exact date of my five-year anniversary at BSF. I believe birthdays and anniversaries have a way of making us reflect on where we've been and where we are now. After the event was over, I thought about what putting on a brave face looked like back when our foundation started sixteen years ago. I imagined our founding families coming together for the first time, wondering if there were any other boys out there like theirs. Many of them have told me how they felt alone and desperate for answers prior to coming together.

Back then, where there was once darkness and fear, an idea was born and hope was sparked. Those parents didn't know what to expect, but they were brave and they searched for answers. Through blood, sweat, and tears, our foundation came to life and began to take root.

More bravery ensued: building a community, providing care, and identifying more boys and men.

Today, we are talking about actual clinical trials for these boys and men. These trials will require our "Barth guys" to be brave yet again. Our boys and men will have to raise their hands to volunteer… to be willing to make some sacrifices to find answers... to help make the future brighter for the younger ones and even for those not yet born.

Bravery is what it took to get to this place where we are now. And, we are here today, on the cusp of such great possibilities, because of all of you. Each has done his or her part, including our donors. Your labor, love, and resources have made it possible for our boys to practice their bravery every day. We need your help to continue to stand with these brave boys and men to get new treatments and, one day, a cure. Thank you for supporting them.

Lindsay B. Groff

Lindsay B. Groff
Executive Director

Building Toward A Miracle

By Marc Sernel, Chairman, Barth Syndrome Foundation

We thought it might never happen. There were a lot of tough times along the way. Even when things were going well, we always worried about when the other shoe would drop and things would turn for the worse. Along the long journey, we lost loved ones who had dreamed of when this day would come but did not live to see it happen. Excitement built this year that maybe this was our time. Then it looked like we would fall short again. And then everything went right and it happened. It was a long time coming. We mourn those who didn't live to see the day. But "we" finally did it.

The Chicago Cubs won the World Series. As a life-long Chicagoan and Cubs fan, it's something I never thought I would be able to write or say. For those of you who are not from the United States or baseball fans, let me try to put this into context. (For those of you in the UK, the Leicester City miracle in 2016 is different but a close enough proxy.) The longest-running drought in the history of American sports was broken in November. The Chicago Cubs had gone 108 years without winning the championship. They had come close during the last century, but something had always gone wrong. Many Cubs fans lived their entire lifetime and never saw the Cubs win. When the Cubs finally won the World Series many fans were overcome with emotion thinking about the loved ones who didn't live to see it, and the team allowed fans to write the names of the deceased on the stadium walls in tribute.

Pardon my analogizing the life-threatening struggle of Barth syndrome with the much-less-important perils of sports — as a Barth parent I certainly understand the difference — but I look forward to BSF also finally finding success and joy at the end of our long and hard journey. Just like the Cubs, BSF has been building a team to put success — for us, a life-saving treatment for Barth syndrome — within our reach. We have certainly faced our share of disappointment and grief, and our hope has sometimes been crushed with despair. We might need some luck, like the Cubs, to finally reverse our fortunes. But I have faith that, with our continued efforts, the future will not follow the script of the past. Many once-fatal diseases have become manageable due to modern medicine, and it is our goal to add Barth syndrome to this list! Miracles in sports, and medicine, do happen!

B.J. Develle

Nicole Derusha-Mackey

I am pleased to announce that we have added two passionate and devoted new members to our BSF Board of Directors to help make the miracle a reality. BJ Develle and Nicole Derusha-Mackey are long-time volunteers who are known to many in our community, and we are very excited to add their talents and perspectives to the Board. BJ has actually been with BSF a lot longer than I have, taking time off from his job as a social worker to be omnipresent at our biennial conferences, working with our young people and willing to help in any other way he can. Nicole has also been very active with BSF for a long time, since 2008. She is the mother of two boys born with Barth syndrome: Nathaniel, who passed away at just two weeks of age in 2002, and Devin, who is now 12. In addition to the great assistance she's already been providing to BSF in terms of outreach and social media, Nicole also brings to the Board her unique perspective of raising a Barth boy after heart transplant surgery. Welcome, BJ and Nicole!

We sometimes take our sports teams and their performance too seriously, maybe because they are a microcosm for life itself. Those affected by Barth syndrome can't really partake in sports, but they feel more acutely the ups and downs of life itself. Some kids hope to grow up to be a major leaguer someday; Barth kids hope they can just grow up. But we are determined to change this, to give Barth kids the ability to live and dream and compete just like any other kid. I thank you for your support, and look forward to celebrating with all of you someday when we win our fight with Barth syndrome. *(Photos courtesy of Amanda Clark [2016])*

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2016 Science and Medicine Sessions
A Glimpse of the Final Frontier?

(Cont'd from page 1)

Pathomechanisms(s) of Barth Syndrome

The Thursday morning session focused on the **pathology of Barth syndrome** (BTHS). Dr. William Pu (Boston Children's Hospital) led off the session by speaking about his work with induced pluripotent stem cells and the abnormalities he found with reactive oxygen species and calcium handling. Dr. Adam Chicco (Colorado State University) used the knockdown mouse model of Barth syndrome to show how the BTHS heart is different from other cases of pediatric heart failure. Dr. Colin Phoon (New York University School of Medicine) continued the theme by using the mouse model to show how the cardiomyopathy in this animal model resembles that found in humans with BTHS. Dr. Saskia Wortmann-Hagemann (Paracelsus Medical Center) spoke about how other organic acid dysfunctions resemble BTHS, while Dr. Laura Cole (University of Manitoba) presented her data about the fat dysfunction in the knockdown mouse model. Dr. Douglas Strathdee (Beatson Institute) described his new knockout mouse model of BTHS and the reproductive difficulties encountered in making this mouse strain.

Dr. William Pu

Dr. Adam Chicco

Dr. Colin Phoon

Dr. Saskia Wortmann-Hagemann

Dr. Laura Cole

Dr. Douglas Strathdee

Ron Bartek
(Photo courtesy of Tiffini Allen [2016])

Keynote Address

At noon, Ron Bartek of the Friedreich's Ataxia Research Alliance gave an inspirational and emotional **Keynote Address** about how his organization (and now BSF) is reaching this new "clinical treatment era." Ron is a leader in the rare disease patient community. His organization is traveling the path that BSF is now just starting upon. Ron spoke about their experiences, and he provided encouragement and support to the entire BSF community at this critical time in its history.

Potential Therapies for Barth Syndrome

The Thursday afternoon session focused on **clinical trials and therapeutic ideas**. Leading off the session was Dr. Todd Cade (Washington University School of Medicine) who spoke about the value of exercise in treating BTHS. Dr. Yuguang (Roger) Shi (University of Texas Health Science Center at San Antonio) showed how disruption of the ALCAT1 gene, an enzyme that interacts with cardiolipin, is able to suppress the cardiomyopathy in the knockdown mouse model implying that inhibition of this enzyme may be therapeutic. Dr. Michael Chin (University of Washington) brought the audience up to date on his progress with enzyme replacement therapy for BTHS. Both Dr. Zaza Khuchua (Cincinnati Children's Hospital Medical Center) and Dr. Mindong Ren (New York University School of Medicine) spoke about the effects of the common drug, bezafibrate, on cardiomyopathy in the knockdown mouse model. Dr. Christina Pacak (University of Florida) revealed a plan of how gene therapy will be used for treating BTHS individuals and the types of delivery vectors they will use. Dr. Brittany DeCroes Hornby (Kennedy Krieger Institute) filled in for Dr. Hilary Vernon and described their study of BTHS individuals at previous conferences and at the Kennedy Krieger Institute Barth syndrome clinic. Dr. Hornby described those clinical characteristics that will be important in performing any clinical trial for BTHS.

(Photos of physicians courtesy of each physician [2016])

(Cont'd on page 5)

Dr. Todd Cade

Dr. Yuguang Shi

Dr. Michael Chin

Dr. Zaza Khuchua

Dr. Mindong Ren

Dr. Christina Pacak

Dr. Brittany DeCroes Hornby

2016 Science and Medicine Sessions
A Glimpse of the Final Frontier?

(Cont'd from page 4)

Poster Session

A well-attended **poster session** where 30 posters were presented followed the Thursday afternoon session. Four poster presenters were selected to speak about their work on Friday. This poster session was open to all conference attendees and was enthusiastically received, especially by the families who were able to personally interact with the authors, other researchers, and physicians.

Clinical Characteristics of Barth Syndrome

On Friday morning we heard more details about the **clinical characteristics of Barth syndrome**. Dr. Colin Steward (Bristol Royal Hospital for Children) focused on the neutropenia aspects of this disease and the implications for proper management of this often overlooked symptom. Dr. Cynthia James (Johns Hopkins School of Medicine) described her efforts to study the social and psychological impact of carrier females in the BSF community. Dr. Brittany DeCroes Hornby (Kennedy Krieger Institute) spoke about the physical strength decreases she measured at the 2014 conference. Dr. John Jefferies (Cincinnati Hospital Children's Medical Center) explained the clinical cardiovascular aspects of this disease. Two poster presenters also talked about their work. Nikita Ikon, a graduate student at the University of California at Berkeley and Children's Hospital Oakland Research Institute, spoke about the nanodisk technology being used for lipid replacement therapy, and Dr. Markus Keller (Medical University of Innsbruck) spoke about his work dealing with mathematical modeling of lipid data.

Prof. Colin Steward

Dr. Cynthia James

Dr. Brittany DeCroes Hornby

Dr. John Jefferies

Nikita Ikon

Markus Keller

John (age 34) presents Varner Award to Dr. Michael Schlame
(Photo courtesy of Tiffini Allen [2016])

Varner Award for Pioneers in Science and Medicine

At noon on Friday the **Varner Award for Pioneers in Science and Medicine** was presented to Dr. Michael Schlame (New York University School of Medicine) who currently chairs BSF's international Scientific and Medical Advisory Board. True to his modest nature, Dr. Schlame spent a minimal amount of time on stage after thanking his colleagues at New York University and BSF for making this conference so special — the most meaningful meeting that he attends.

Cardiolipin and Barth Syndrome

On Friday afternoon the **connections of cardiolipin to BTHS** were explored in more detail. Dr. Robin Duncan (University of Waterloo) produced her data showing that there may be a new cardiolipin synthesis pathway. Dr. Valerian Kagan (University of Pittsburgh) showed that cardiolipins can alter intra and extra-mitochondrial events with their vast potential of different forms which can serve as signaling molecules. Dr. Miriam Greenberg (Wayne State University) related how metabolism in cardiolipin deficient cells perturbs all three major components of energy metabolism. Dr. Nathan Alder (University of Connecticut) showed his results about modeling of cardiolipin with membranes and membrane proteins including changes in its ionization properties. Two poster presenters also spoke about their work. Dr. Gang Wang (Boston Children's Hospital) showed that the cardiomyopathy of the knockdown mouse model is genetically reversible and can also be reversed with the Bendavia compound. Dr. Anthony Aiudi of Stealth Biotherapeutics discussed the current pre-clinical and clinical results of the Elamipretide/Bendavia compound on mitochondrial and other diseases.

(Cont'd on page 6)

Dr. Robin Duncan

Dr. Valerian Kagan

Dr. Miriam Greenberg

Dr. Nathan Alder

Gang Wang

Anthony Aiudi

2016 Science and Medicine Sessions
A Glimpse of the Final Frontier?

(Cont'd from page 5)

Conference Wrap-Up

The four conference chairs of the SciMed sessions, Drs. Arnie Strauss (Cincinnati Children's Research Foundation), Todd Cade (Washington University), Colin Steward (Bristol Royal Hospital for Children), and Ronald Wanders (University of Amsterdam) provided **summary remarks** about the SciMed sessions. Besides listing the important advances and new ideas that were discussed over the preceding two days, these BSF-sponsored conferences were found to be remarkable for the openness and frank discussions of unpublished material, which has helped to stimulate the real progress seen at this meeting.

Friday evening featured a **social event** where all conference participants got together to have a good time and to celebrate the amazing advances. Saturday morning allowed the **Scientific and Medical Advisory Board** to meet where they heard a presentation by Stealth Biotherapeutics and their plans for an upcoming clinical trial with Barth syndrome individuals. On Saturday at noon the finale of the conference was held.

It was a spectacular conference from many perspectives, and the new friendships gained and the old ones renewed will keep propelling BSF toward its goal now and for years to come. Undoubtedly it was a pivotal moment in the history of BSF; one where the community could glimpse the "final frontier" in its quest for the ultimate goal. *(Photos courtesy of BSF 2016)*

(L-R) Dr. Brittany DeCroes Hornby, Rebecca McClellan & Dr. Yana Sandlers

(L-R) Dr. Angela Corcelli, Dr. Michele Dibattista & Dr. Simona Lobasso

(L-R) Dr. Colin Phoon, Dr. David Kronn & Dr. Mindong Ren

Team Barth!

Doctor dance off!

Dr. Carolyn Taylor, R.J. (age 18), Dr. Barry Byrne & Dr. Todd Cade

Barth Syndrome Researcher Awarded R01 Grant from the National Institute for General Medical Sciences

Dr. Michael Chin

Michael Chin MD, PhD, Associate Professor, University of Washington, Seattle, WA, was awarded an R01 grant from the National Institute for General Medical Sciences for his proposal entitled "Intracellular mitochondrial enzyme replacement therapy for heart and skeletal muscle in Barth syndrome." This grant will enable Dr. Chin to continue the work that he began with initial funding from the Barth Syndrome Foundation. He will study the mechanism by which recombinant *tafazzin* enters the cells and travels to the mitochondria, measure the pharmacokinetics and tissue distribution of the enzyme replacement therapy, and measure effectiveness of the enzyme replacement therapy in correcting cardiomyopathy, skeletal myopathy and neutropenia in a mouse model of Barth syndrome. *(Photo courtesy of Dr. Michael Chin 2016)*

Our Brand New Threshold of Hope — Clinical Trials

(Cont'd from page 1)

But we cannot move forward without your help. If we do not embrace this golden opportunity now, we will lose it.

Those of you who attended this session for families at the conference heard a lot about the therapies and what participating in a clinical trial actually means. With Kate's permission, I want to review some of the information she shared with us. If you were not at the conference, this article will provide you with basic information about these incredible developments. I strongly encourage you to listen to Kate's part of the session which has been posted on BSF's YouTube channel. It is called the "New Frontier" Family Sessions. I'd like for each of you to hear this first-hand from Kate, as it is critical that we all learn as much as we can about clinical trials and what it means to be a part of one. What I have written below is a VERY abbreviated version of what we learned in this session.

Kate first discussed the steps and stages of advancing a therapy through the clinical development process. Here in the United States, this process is under the direction of the Food and Drug Administration (FDA). Each country has its own agency and regulations. The FDA requires that all potential new treatments be tested in clinical trials before they are reviewed for approval for use in the US. The next topic was what it means to participate in a clinical trial; I have summarized this information below.

First of all, what is a clinical trial? According to the National Institutes of Health, clinical trials are research studies that explore whether a medical strategy, treatment, or device is safe and effective for humans. These studies also may show which medical approaches work best for certain illnesses or groups of people. Clinical trials are carefully designed and involve human volunteers. The FDA is especially focused on the safety and effectiveness of new therapies, and they have set up very strict guidelines to this end.

What do clinical trials have to do with me and/or my family? **Volunteering to participate in clinical trials directly affects all who have Barth syndrome or who love someone with Barth syndrome. In short, THIS IS OUR MOMENT. As Kate explained, this is THE way that we get treatments for Barth syndrome. Science will not go forward if people do not sign up to participate, and our chance will not come around again.** 80% of clinical trials fail to recruit enough volunteers within the time schedule that was planned — in fact, not recruiting enough volunteers is the leading reason why clinical trials cannot be conducted. Our challenge is that if we cannot get enough people to participate in the upcoming trials (and we hope there will be a number of them in the coming years), we will never get another chance with that particular drug. Drug companies do not come back around to try again.

One thing that makes our situation more difficult is that many clinical trials have hundreds, or even thousands, of people willing to participate. With a rare disease like Barth syndrome, we have a small number of potential volunteers because so few people have the disease. We currently know of around 200 people with a confirmed diagnosis of Barth syndrome worldwide, so it is critical that people in our community who meet the criteria for each study really consider participating in a clinical trial. Our community is particularly attractive to the people conducting trials because we are strong and committed, and we are the largest group of people with Barth syndrome in the world.

I am interested in helping. What's next? BSF will notify our community when a clinical trial is going to begin. Information will be distributed about the trial, such as what the study involves, who is eligible to participate, and who cannot participate (perhaps people with a heart transplant or an ICD, or younger than a certain age, for example). This information is part of the study protocol. Also, Shelley Bowen, Director, Family Services and Awareness and Matt Toth, BSF Science Director plan to hold webinars with the PI (Principal Investigator) for each trial as trials come up.

What is a study protocol? The study protocol is the plan that describes in detail how the clinical trial will be conducted. Very simply, this is the who, what, when, where, why and how of the study. It is carefully designed to safeguard as well as possible the health of participants and to answer specific research questions. As noted above, the protocol describes who may participate in the trial, what tests, procedures, medications may be required, the length of the study (the study needs to be long enough to get results, know that the medication is safe, and compare results), and what travel is required for volunteers as well as other information related to the trial.

Then what? Once volunteers who meet the criteria have agreed to participate in the trial, the **informed consent process** begins. Many of you are already well versed in this process, but this is where a member of the study team will explain everything that is involved in being a study participant. If you decide you are interested and you meet the eligibility criteria, you will be given a thorough explanation of the study protocol before you officially agree to participate. You will also be told how the results will be determined, and possible risks and benefits of the study will be explained in detail.

Can I ask questions during the informed consent process? It is important for you to ask every question you can think of during this process and at any time during the study — there are no silly questions. Some things you might ask include: Who has approved this study? Why is this a good idea? Who is in charge if I have a question? What happens if I have a problem or if I get sick? What should I watch for? What kinds of problems might I see? What are the hoped-for results? Will taking my current medications (including amino acids) be allowed during the study?

Other questions you might want to ask during the informed consent process are: Who pays for everything? Will my insurance be involved in paying for any of the tests? If so, how will this be handled? How much travel will be involved and for how long? Can

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Our Brand New Threshold of Hope — Clinical Trials

(Cont'd from page 7)

a parent attend out of town appointments with the participant, and if so, would that be paid for? Would lodging be paid for? Please give this lots of thought — make a list of your questions, and do not hesitate to ask anything.

What are the benefits of participating in a clinical trial? Benefits can include having an active role in your or your child's health, gaining access to a new treatment before anyone else, receiving expert medical care, and advancing medical research for yourself and other people affected by Barth syndrome.

What are the risks? There are potential risks involved with nearly every single thing we do: we always have to weigh the benefits of anything we do with possible risks. Some potential risks of participating in a clinical trial are that the drug may not be effective, or that the protocol may involve more time, attention, discomfort, or travel than we are used to. There could be potentially serious or life-threatening side effects. It is possible that being a part of any study could exclude you from participating in a different study in the future, depending on the criteria.

But, as Kate pointed out in her talk, and I agree with 100%, there are also side effects and risks to doing nothing. Not only might you miss out on getting a potential treatment early, but if too many people choose not to participate because they are concerned about these possible risks, then all individuals affected by Barth syndrome miss out on potential life-saving treatments. The researchers in charge of each study will carefully review the potential risks with each participant during the informed consent process. Again, please **ask any and all questions** when the risks are being discussed.

Additionally…. It's very important to keep your local doctors in the loop if you participate in any of the clinical trials for Barth syndrome. They will need to know that you are a part of a trial and what it involves, that you are on a new medication, what their responsibilities may be, and whom they can contact if they have questions about the trial.

Participation in any clinical trial is completely voluntary. If you do agree to participate and change your mind before or during the study, you can stop at any time, for any reason.

More information about future clinical trials will be coming. Wonderfully, it looks like there will be a number of trials, all with different protocols. We need volunteers to sign up for every one of these trials. If you cannot sign up for the first one or two, please know there will be other opportunities!

It is important to understand that not every trial will work. Some will not. The only way we will find out which trials can lead to treatments for Barth syndrome is to TRY. The only way that we can try is for people with Barth syndrome to participate in clinical trials. No one else can do this for us. If the first few trials are not successful, it's critical that we do not give up. We cannot get discouraged and quit, because this is the only way we will ever be successful in finding treatments for Barth syndrome.

We are truly standing on a brand new threshold right now — one we've worked toward for a very long time. It represents hope. It represents our future. We have suffered through profound heartbreak because of not having treatments available for Barth syndrome. We have lost too many babies, boys and young men whom we love to this disorder. Let's gather our extraordinary strength as members of Team Barth and be a part of our collective future, making our BSF vision "A world in which Barth syndrome no longer causes suffering or loss of life" our new reality. Let's *Grow Stronger* together.

(Some information used in this article is from TS Alliance educational material.)

William (age 12) and his mom, Julie, having blood drawn

Cameron (age 17) having vitals taken by Sunstar paramedics
(Photos courtesy of Tiffini Allen [2016])

Opportunities to Participate in Barth Syndrome Research

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.)*

Heart and Skeletal Muscle Metabolism, Energetics and Function in Barth Syndrome

W. Todd Cade, PT, PhD, has received notice of an R01award from the National Heart, Lung and Blood Institute of the National Institutes of Health for his project. This award is distributed over five years and is in the amount of US $1.6 M. This is a significant achievement for which BSF is very proud and excited. Dr. Cade began recruitment for this research project at BSF's 2012 Conference! *(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).

Barth Syndrome Foundation does not endorse any drugs, tests, or treatments that we may report.

First-Ever Pill-Swallowing Clinic a Success at Barth Syndrome 2016 Conference

By Stacey Reynolds, PhD, OTR/L and Emily Burgess, MS, OTR/L, Virginia Commonwealth University, Richmond, Virginia

"The Pill-Swallowing Clinic has made a world of difference for me. Instead of enduring pain, I can now actually swallow medication — this is huge for me." *~ Adam, Affected Individual (age 26), Ontario, Canada*

For children and adults with chronic health conditions, taking medications orally is part of their everyday lives. Swallowing pills, however, can be a challenge to individuals whose diagnostic condition includes sensory sensitivities, postural control deficits, and oral-motor coordination problems. This is the case for many individuals with Barth syndrome.

Dr. Stacey Reynolds

Why is it important for individuals with Barth syndrome to be able to swallow pills? While some medications may come in liquid format, oftentimes pill format is the only option. Liquid medications can likewise be a hassle for individuals and families to manage while traveling due to the need for medication to be kept at a specific temperature. Also, there are new clinical trials being initiated for individuals with Barth syndrome; however inclusion in the trials often requires participants to have the ability to swallow large encapsulated pills. Overall, the ability to be independent in swallowing medication in pill format is an important self-care milestone for many individuals with Barth syndrome, but a milestone which may be challenging for a variety of reasons.

To help overcome challenges to pill swallowing, our team from Virginia Commonwealth University was invited to lead a Pill-Swallowing Clinic at the 2016 International Barth Syndrome Conference held in Clearwater Beach, Florida. Before running the clinic, our team went to the literature to see what had worked before with other populations of children and adults. The most common approaches to pill-swallowing employed a technique called behavioral shaping in which individuals practiced swallowing a variety of candy that progressively increased in size (*e.g.,* cupcake sprinkles, Nerds, tic-tacs, M&M's, and Mike & Ikes). Other strategies included altering head positions during the swallow (*e.g.,* with/without chin tuck, or head turned to side) and use of adaptive equipment like the Oralflo pill swallowing cup (http://oralflo.com/) and flavored Pill Glide sprays.

Andrew (age 27) during Pill-Swallowing Clinic
(Photos courtesy of Kevin Boozer [2016])

At the clinic, questionnaires and clinical tools were used to evaluate sensory, motor, cognitive and behavioral/psychological issues related to pill swallowing. Based on the participant's age and profile, we selected evidence-based strategies to help facilitate pill swallowing. We assessed progress based on the subject's willingness to engage in the pill-swallowing process and the size of pill (candy) they were able to swallow. Out of the 16 boys and men who participated in the clinic, all 16 made significant progress towards pill-swallowing independence and many were able to progress to the largest size pill (candy) by the end of the brief intervention.

Interestingly, the techniques that worked best for younger children differed from those that worked for older adolescents and adults. In general, children who had never before tried swallowing a pill (*i.e.,* no previous exposure to pill swallowing) were able to use a regular cup with water and only a slight tip-back of the head in order to swallow the candies. These boys often found the challenge of increasing candy size motivating, and enjoyed practicing a new skill in a stress-free environment. However, older males with Barth syndrome who did have prior pill-swallowing experience often entered the clinic with some anxiety about pill swallowing and trepidation about working on this skill. For these individuals, the Oralflo cup was often the most successful tool to pill-swallowing independence; this is likely because the individual does not see the pill prior to swallowing and just drinks from the cup's spout without having to first place the pill on their tongue. Some older boys and men also found the alternative head positions useful (*i.e.,* head to left or right) for swallowing, particularly if they had prior experience of having the pill get caught in their throat.

Overall, we received positive feedback about the pill-swallowing clinic and were impressed at the progress each participant made. Our take-home message to Barth families is that pill-swallowing should be practiced in a stress-free setting and should take into consideration the sensory, motor, and behavioral needs of each individual. We hope to provide the clinic at the 2018 conference. In the meantime, questions can be addressed to reynoldsse3@vcu.edu or burgessme@mymail.vcu.edu.

Team Barth: 2016 Barth Syndrome Conference United in Laughter, Blood, Sweat and Tears

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

"For years I wondered if my research would ever make a difference to a human being and then I came to this conference and there they were. And that has made a tremendous difference to me personally and to me on a professional level." *Miriam Greenberg, PhD, Wayne State University, Detroit, Michigan*

Over the years, technological advances have made it possible for our families to bridge geographical barriers when they are in need of help. But there is no app that can replicate being in the presence of others who just "get it." Our conference every two years brings together those who get it and experts who are doing something about Barth syndrome (BTHS).

In one short week, we do it all. Families participate in research, learn from each other and hear about progress that has been made in research. Scientists have the opportunity to (1) learn about the latest scientific developments and clinical insights into Barth syndrome; (2) exchange ideas with others involved in the many aspects of this complex disorder; (3) help accelerate advances in the understanding and treatment of Barth syndrome and shape the future of Barth syndrome research; and (4) network with individuals personally affected and their families. Clinicians have the opportunity to gain information about symptoms and latest research findings and have access to other members of the medical community worldwide with experience of diagnosing and treating Barth syndrome.

Dr. Brian Feingold
(Photo courtesy of Tiffini Allen [2016])

Scientists who seldom meet the people who benefit from their life's work have told us that attending our conference has been transformational. We set aside time to make memories, and we set aside time to remember all those who have ever lived with Barth syndrome. Families travel from all corners of the globe to unite in laughter, blood, sweat and tears. For one week, researchers and relatives of those who have Barth syndrome become one family that lives on very little sleep. Some people are amazed at the length of our event, thinking it's just too long. However, most would agree it's not long enough.

We don't just hope collaborative discussions will transpire between scientists, we facilitate opportunities for these to take place. Our scientific and medical attendees find our model a refreshing change from most meetings they attend. The conference experience is difficult to describe. It's something one must experience to appreciate. When a clinician who treats only a few patients with Barth syndrome, such as Dr. Brian Feingold, says, ***"This is my first conference, but it won't be my last,"*** we know he has become a member of Team Barth.

The conference is more than a series of talks. On average, approximately one quarter of the world's population of affected males who have Barth syndrome attend our conference. Affected males, family and friends participate in clinical research. This year, 168 people participated in seven IRB-approved studies for a total of 660 appointments scheduled over a 3-day period. There are many reasons why investigators prefer doing research during the conference. For us, it's simple — it's the smart thing to do. We estimate the cost savings from the 2016 conference to be approximately $520,000. Researchers come to the families rather than the families traveling to the research center. This alone is a cost savings of nearly $240,000. Often times, BTHS investigators donate their time and pay their own costs for travel and lodging. Institutional research collaborations and donations of materials from corporate sponsors cover material costs and save tens of thousands of dollars. The data collected during this conference will provide insights about the clinical aspects of BTHS. Results from these studies will be published in journal articles, which will expand the base of knowledge about this rare disease.

Cost Savings

Travel	$ 237,750
Lodging	$ 59,235
Stipends	$ 41,200
Materials	$ 179,510
TOTAL VALUE	$ 518,495

(Cont'd on page 12)

Team Barth: 2016 Barth Syndrome Conference United in Laughter, Blood, Sweat and Tears

(Cont'd from page 11)

"Attending the conference was the best experience of our lives. Meeting so many people from so many walks of life united by the same common cause was like being with people who fully understood us for the first time since my son was born. " *~ Jason, Father of Affected Individual (age 5), Illinois*

The theme for this year's conference was "Team Barth." Multidisciplinary experts who have investigated boys and men with Barth syndrome (BTHS) led workshops to provide families with practical strategies they so need to help them on a day-to-day basis. We are now entering an exciting phase of developing therapies through clinical trial research studies. We reached out to occupational therapy specialists, Dr. Stacey Reynolds and Emily Burgess, to ask them to lead a pill-swallowing clinic. As an investigator who has led numerous BTHS research studies since 2010, Stacey was very familiar with the challenges those who have BTHS experience with swallowing. This simple inconvenience could possibly have been a great barrier in the clinical trial studies where participants are required to swallow a pill. The one-on-one coaching to learn swallowing techniques and tools to help with swallowing was a tremendous success with all participants (age 5 – 36 years of age). Conquering this hurdle is a remarkable feat for many of our boys and young men.

(L-R) Nicholas (age 18) & Jack (age 23)

One of the greatest testimonials of impact of our mission is that many of the boys who first came to us in the early years of BSF are now older. New challenges emerge as these boys grow up, so our dream-team of experts led independence and transitions educational tracks. These sessions were divided into age-specific topics to address needs of our families and provide practical resources developmentally appropriate to age. They included tips and insights about nutrition, adaptive technology, building relationships with healthcare professionals, relationships with peers, education and how to talk with your child about having BTHS or potentially being a carrier of Barth syndrome. This year we continued our carrier discussions and heard updates about the carrier research study that arose as a result of the needs expressed during our 2014 conference.

Our mission has always been the compass to guide us. When Kate McCurdy pointed to the midpoint of a clinical trial roadmap during her presentation to the families, our families gained a clear view of the progress we have made since 2000. There is more ground to cover, but the verve in the room strengthened as researcher after researcher presented opportunities about potential therapies for BTHS. No longer is the brass ring of Barth syndrome therapies a distant dream but is realistically within our grasp. We have always had hope that we would someday reach our ultimate vision of ending suffering and death caused by Barth syndrome. With clinical trials in the pipeline, that hope may well become a reality in the not-so-distant future. Every attendee, regardless of role or relationship who had ever attended a previous conference hailed the 2016 conference as the best one ever. Every first-time family attendee overwhelmingly found the conference to be transformational.

Many of our Barth boys and men, along with their siblings, at BSF's 2016 conference *(Photo courtesy of Amanda Clark 2016)*

BSF Brings Comfort, Support, Good Times, Hope and Happiness

By David, Father of Affected Individual, Tennessee

(L-R) David & Ben (age 19)

My name is David, and I am 49. I have two wonderful children, English and Ben. English is 23 and Ben is 19 and has Barth syndrome. I also have one lovely wife, Shelia. We just celebrated our 25th wedding anniversary! We live in Knoxville, Tennessee.

We have been very blessed to attend every Barth conference since the beginning in 2000. Ben was only three years old and English was seven when it all started in Baltimore, Maryland. There, for the first time, we met other families with the same fears, concerns and questions that we had. Seeing another small boy with Barth syndrome just like our Ben for the first time was overwhelming.

As Ben and English grew older, each conference seemed to take on new meaning for us as a family. We experienced new emotions, concerns and milestones over the years. We began to have more hope for a longer life for our son Benjamin. Each conference helped me, as a father, understand and cope with the day-to-day life of having a son with Barth syndrome.

Ben and English, as well as Shelia and I, grew to love each Barth boy, their families and every person involved with the Barth Syndrome Foundation (BSF) with every passing year. We still get very excited about seeing all of our friends, old ones and new ones. The bonds and connections we have made with other families have forever changed our lives. We always look forward to meeting new families each conference and sharing with them how we have navigated the maze of medical and emotional challenges that come along with having a child with a disability.

The beauty of the Barth Syndrome Foundation lies in the fact that no matter where our family is, whether in sickness or in health, there is comfort, support, good times, and happiness to be found. The foundation extends its caring arms not only to the affected boys, but to siblings, moms, dads, grandparents, and cousins. There is something for each of us in this magnificent foundation we call family, and we are fighting together to make a difference.

The foundation means many things to me. HOPE…..We now have HOPE for our son and all those affected by Barth syndrome, plus hope for our carriers and our newly formed Barth family! There's a lot of emotion as well as fun filled good times. It's actually very difficult to describe in a few words. When I am together with the Barth dads, the good times seem to roll. Fears and concerns seem to soften; however, they never go away.

I am very proud to see our son and all the guys stepping up at each conference to participate in research and mentor the younger boys! I have often wondered if all the efforts our boys and young men have put forth will make a difference. We are now starting to see they have made a difference. New treatments are in the very near future for our son and all the guys. I am very excited about the promising future for our boys. There are a lot of big decisions coming in the near future with the exciting new opportunities for gene therapy and other treatments.

It's hard to believe our son and family have been involved with BSF for 16 years now. We just this year had to transition Benjamin into adult cardiology care, a milestone we never thought we would see. This is an amazing gift that BSF and all of Ben's doctors and caregivers helped our family achieve, and we are forever grateful.

It's always tough to say goodbye at the end of each conference, given that it will be a long two years before we see each other again, but we always pick up where we left off!

(L-) Ben & David in Italy
(Photos courtesy of David 2016)

See you in 2018!

You Make a Difference

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

Thanks to all of you, we continue to serve families affected by Barth syndrome and fund groundbreaking research. From breaking boards in the name of Barth syndrome to running marathons, you've made a difference. Thank you! We couldn't do it without your help.

2016 Conference Sponsors

So many of you donated to BSF to ensure that our 2016 Conference was a success. It sure was, thanks to your support. As you can read in this newsletter, this may have been our best conference yet. Your donations made it possible to pull off such a huge undertaking. We are grateful to each and every one of you. A very special thanks to our donors, our sponsors, and our volunteers. We couldn't do it without you! *(Photo courtesy of BSF 2016)*

(L-R) Dilen, Milosh (age 6) & Ronly display sponsor sign

Touw family

Community-Wide Yard Sale

Along with members of her neighborhood, Lindsay Groff held her annual community-wide yard sale with proceeds given to the Barth Syndrome Foundation (BSF). Over a dozen houses sold household items in all. The kids in the Touw family also held a bake sale and donated all that they earned to BSF. *(Photo courtesy of Lindsay Groff 2016)*

Michael Neece

Breaking Barth 2016

Neill's Family Taekwondo is committed to breaking Barth syndrome by raising awareness through fundraising and breaking boards as a symbol of our desire to smash this disease. Over the past two years, they've broken first 500, then 1,000 boards, raising over $4,000 so far. This year, instead of a single board-breaking event, we are issuing the Board Break Challenge (much like the famous Ice Bucket Challenge). Friends will challenge each other, their families, teachers, and students to break a board in a short video via social media, and make a donation to the Barth Syndrome Foundation. *(Photo courtesy of Michael Neece 2016)*

Eleanor & Eliza

Navy Air Force Half Marathon

On September 18, 2016, Eleanor Fanto ran for Team Will in the Navy Air Force Half Marathon & Navy 5 Miler in Washington, DC. When asked what inspired her to participate in this event, she said, **"Eliza McCurdy is one of my best friends in the world. We met in nursery school, went to high school together, and now we're both rising seniors at Georgetown University. I have always loved her parents and Will very much. Therefore, I'm incredibly excited to challenge myself athletically and fundraise for Barth syndrome at the same time by running this half marathon."** *(Photo courtesy of McCurdy family 2016)*

Members of Team Will

Westchester Olympic Marathon

On September 25, 2016, Heather Segal, Laura Azar Fairchild, Stefan Tunguz and Francois Odouard, members of Team Will, participated in the Westchester Olympic Marathon for the tenth year! As Heather wrote, **"For the past 10 years, I've competed in triathlons as part of Team Will to raise money to support the Barth Syndrome Foundation (BSF). Huge thanks to those of you who have generously supported BSF in the past. Will's passing has motivated me to try to raise even more funds to help find a cure for this dreadful disease. Your donations help…a lot! BSF expects to begin two different trials of promising compounds to treat Barth syndrome — likely within the next six months. Yet, the BSF community still needs our support to help drive this progress forward."** *(Photo courtesy of Heather Segal 2016)*

(Cont'd on page 15)

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Toniolo, Dr. Daniela
Toth, Dr. Matthew J.
Towbin, Dr. Jeffrey A.
Tunguz, Stefan, Leigh & Julie Kinch
Van der Laan, Dr. Martin
van Loo, Joke
Vance, Dr. Jean E.
Vartak, Dr. Rasika
Venditti, Dr. Charles P.
Veritus Group
Vernon, Dr. Hilary
Vicart, Dr. Patrick
Wanders, Dr. Ronald J.A.
Wang, Dr. Hay-Yan J.
Weber, Dr. Thomas
Whitney Media
Wilkins, John & Sue
Witt, Dr. Martin
Witt, Dr. Stephan
Woodward, Kevin & Stacey
Wortmann-Hagemann, Dr. Saskia
Wright, Jessica
Ye, Cunqi
Yin, Dr. Huiyong
Yu, ScD, Xin
Zaragoza, Dr. Michael V.
Zeller, Anne
Zhang, Dr. Ji
Zhang, Jun

You Make a Difference

(Cont'd from page 14)

Cambridge Half Marathon

Russell and Ali Pierson are Will McCurdy's first cousins. They grew up together, sharing summers in Lakeside, MI (Will's final resting place) and many visits to each other's homes for graduations, birthdays and holidays. They witnessed first hand the challenges that Will faced every day and the courage with which he lived his life. Will served as an example for Ali and Russell and all who knew him of how to keep your focus on what is truly important and never give up. In return, Will drew his strength from their love for him. On November 13th, Russell and Ali entered in the Cambridge Half Marathon outside of Boston and dedicated their race to the memory of their cousin, Will. Like all members of Team Will, they knew that Will's example would sustain them during the toughest parts of their race. Your donations in their honor helped BSF bring treatments to Will's Barth "brothers" and ultimately help find a permanent cure for this devastating genetic condition. *(Photo courtesy of Russell & Ali Pearson 2016)*

Russell & Ali Pearson

New York Islanders Game

Wyatt did the puck drop and appeared on the Jumbotron

Just as we were going to press with this issue, the New York Islanders held a "Barth Syndrome Night" and allowed us to raise awareness and money for our cause. Nine-year-old, Wyatt, appeared on the Jumbotron, rode the Zamboni, high-fived all the players, got an official jersey, and even did the puck drop! More to come on this exciting night in the next issue! *(Left photo courtesy of New York Islanders; right photo courtesy of BSF 2016)*

Volunteers at NY Islander Game

In Loving Memory

Beverly Lever & Shelley Bowen

Beverly Lever died on June 2, 2016. Beverly was the mother of Shelley Bowen and grandmother of Shelley's children, Michael, Evan and Alanna. Beverly brought joy to everyone she met, sharing her giving heart and gentle soul. She was a constant encourager in Shelley's life. She went to great lengths to be there for Shelley when her boys were ill. And she was there when BSF was started. On November 13, 2015, she considered it her greatest gift to celebrate her 77th birthday with many of the people from BSF she had come to know and love over the years. She will be missed. It was Beverly's desire for donations to be made to the Barth Syndrome Foundation in lieu of flowers. *(Photo courtesy of the Bowen family.)*

Mary Baffa & Mary Kate

Mary E. Baffa died suddenly on October 1, 2016. Beloved wife of the late Theodore F. Baffa (Ted), Mary and Ted had four sons: Ted Jr., Frank, Stephen and David and eight grandchildren including Ted Jr. and Rosemary's children, Matthew, Kevin, Jamie, Mary Kate and Anna.

Mary was a loyal friend and devoted to her family. She was instrumental in helping the Barth Syndrome Foundation start its Scientific Research Grant Program by convincing her long-time employer, Leonore Annenberg, to make the inaugural contributions to the program. As she did when her husband Ted died, the family asked that in lieu of flowers, donations be made to the Barth Syndrome Foundation, in honor of her grandson, Kevin Baffa. *(Photo courtesy of the Baffa family)*

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 **135** 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.

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Barth Syndrome Trust — Update from Chair

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

"It was great to meet up with families and hear about up and coming treatment trials and research."

"Very informative, but very informal, which I found to be especially effective."

Nick & Michaela
(Photo courtesy of Amanda Clark [2016])

Much of this edition focuses on the recent International Conference in Florida in July, which was a huge success. It was also the first time I had been to a BSF conference in a long time. In fact, it had been eight years! For many of our families outside the US, getting to the conference is just too expensive, and often the timing falls within school term time. BSF has posted all the talks online which is a wonderful way to at least access the information that is shared at the conference. What is a lot harder to reproduce is the way attending families immerse themselves totally in the experience, making new friends without all the time and effort it usually takes to get to move past acquaintance into true friendship. It's a bit like a full-on summer camp, just with much better beds and food. It remains a unique opportunity to participate in research; a way of investing into finding a treatment for all our boys and men. One day I hope to find a wealthy benefactor who will invest in this unique opportunity to exponentially increase the advances being made in our knowledge of Barth syndrome by helping as many families as possible attend the conference. After all, that has to be one benefit of being such a super rare condition, doesn't it? With fewer than 200 affected families worldwide, wouldn't it be something if we could help each one of them to attend the research and educational sessions offered?

While we wait for our knight in shining armour, we try to use the funding we have to create the most impact, usually by assisting key people to attend the conference so that they can share their knowledge there as well as bring it back to our families via the UK clinics. This year the Barth Syndrome Trust (BST) in the UK and the Barth Syndrome Foundation (BSF) in the US were proud to co-sponsor the travel costs of Dani Goodman (Occupational Therapist), Nicol Clayton (Dietician) and Lucy Buckley (Physiotherapist) all from Bristol, UK. This wonderful team worked incredibly hard throughout the conference week, providing expert and tailored information to families as well as being able to talk to and learn from their colleagues from all around the world.

After all that work, we needed some fun! BST was pleased to sponsor the Friday Night Social where everyone got the chance to let down their hair and relax after a whirlwind week.

Once back home in the UK, we decided to share some of the main information that came out of the conference with our UK families who were not able to attend. We hosted a mini Barth Information Weekend in October in Bristol. This was very well attended, and we focused on explaining more about the potential new avenues of research, clinical trials and other treatments that are currently being pursued by our dedicated doctors and scientists.

None of this would have been possible without the help of our dedicated fundraisers and donors – thank you to each and every one of you.

Families at the Barth Information Weekend *(Photo courtesy of BST [2016])*

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Barth Syndrome Trust — Update from Chair

(Cont'd from page 18)

Dani Goodman

And lastly, a very sad goodbye…from all of us to the wonderful Dani Goodman. Dani has been an incredible asset to all our families, often travelling hundreds of miles to visit us at home or at school to discuss adaptations that makes the lives of those with Barth syndrome more manageable. Dani has been the driving force behind many new programs. She has the unique gift of seeing what needs to be done and making sure she finds a way to make it happen. Her work has made such a positive impact on the lives of our Barth families. We will miss her energy, expertise and enthusiasm, and we wish her well in her new venture. Don't forget your Barth family Dani, we certainly won't forget you! *(Photo courtesy of BST 2016)*

Back row: Wendy, Ieuan, Ralph
Front row: Kate, Aneira, Alex, Isabel and Thelma
(Photo courtesy of Amanda Clark 2016)

From Kate Riseborough

When my nephews, Ieuan and Alex, were diagnosed with Barth syndrome, we had no idea what it was or where to get more information. Along came the Barth Syndrome Trust and a huge helping hand on our way to understanding. Then came information about conferences; due to it being in the USA, it took until this year to finally get us all there.

We weren't sure what to expect but we were welcomed with open arms and lots of hugs! We were assigned a mentor for the week to help us if we needed it. We met ours, Nicole, who I'd spent so much time chatting with on Facebook and it felt like I'd known her for ages. It was fantastic to meet other Barth families and medical teams who are all dedicated to finding a cure. To have clinical teams to perform research on site too made it fascinating (I'm a lab technician) and a worthwhile conference making lots of new friends. Not to mention the dancing at the Friday night social!!

It has inspired us in many ways, and as a result of this, Wendy and I have started running in order to raise money for Barth syndrome. We have our first 5K run in November where we are raising money for the Trust, and we have also just signed up with runDisney in April to complete a 10K and Half Marathon over a weekend!!

Helen Coleman
(Photo courtesy of Amanda Clark 2016)

From Helen Coleman, Trustee and mother of Will (27 years old)

What did I expect from the Conference?

- To put faces to names that I knew
- To meet newly diagnosed families
- To see how differently all the boys and men were affected
- To hear a bit about the current research
- To be HOT outside the hotel!

What I didn't expect:

- For it to be so emotional
- The amount of organisation and the smooth running overall
- So many doctors and physicians
- The sheer number of families and attendees
- The size and variety of topics covered
- Such a great venue
- That I would miss Will so much…part of me was missing!
- Just how much research is going on around the world
- Just HOW hot it was!!

In conclusion, I found the whole Conference experience moving, uplifting, encouraging, fun, interesting, unbelievable, warm, loving, but beyond all of that, hopeful. I came away on Sunday feeling that one day, there will be a cure for this disease that affects our boys and takes too many of them away too soon.

From Gill Amos, mother of Jack (10 years old)

Time during the conference was divided into research studies; small group meetings and lectures from the scientific and medical team for families; lectures and discussions primarily for scientists and clinicians; and age appropriate information sessions for the Barth guys with the option for them to just chill out or do fun and creative things whilst being looked after by lovely dedicated people.

In conclusion, I think the greatest gift was getting an appreciation of how much is going on and seeing the dedication involved. I have

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Barth Syndrome Trust — Update from Chair

(Cont'd from page 19)

a much greater awareness of what could be in store for Jack and that this time before puberty really could be the best health that Jack will enjoy, unless more therapies are tried and tested. We can't see the future, but we know that working as a team and sharing our expertise and experience will help Jack lead a full and creative life.

Jack
(Photo courtesy of Gill [2016])

From Jack (age 10)

My name is Jack and I am 10 years old. I go to Exeter Steiner Academy and I enjoy school. My favourite subjects are land-based education where we get to do wood work, clearing woodland, making fires and being outside.

My favourite foods are roast chicken skin and macaroni cheese with soya sauce; my favourite vegetables are radishes and green cabbage. My favourite fruits are strawberries and raspberries.

My best hobby is reading, especially Famous Five books and Harry Potter. I am also learning to play the cello and have just started piano lessons. In my spare time I like playing magic imagination games with my friends and riding on my quad bike which was given to me by another Barth boy (thanks Nick!).

My best holiday is going camping, especially voice camp which is a singing camp where we play sword fighting in the woods. I'm not really bothered by having Barth syndrome, I just have my life how it is. At the moment when I grow up I would like to be a farmer, get married and have two sons and two daughters.

Barth Syndrome Foundation of Canada President's Report

By Susan Hone, President, Barth Syndrome Foundation of Canada

It seems like it was a long time ago but, in reality, it has only been four months since my family was fortunate enough to attend the 8th Barth Syndrome International Scientific, Medical and Family Conference in Florida. Every time I attend a conference, I am in awe of what this group of families, scientists, doctors, therapists and others in the medical field have managed to accomplish in such a short time. I never dreamed that we would ever be looking at more than one possible treatment at a time. I also never imagined the magnitude of scientists, doctors, therapists and others from around the world who have become dedicated towards the Barth syndrome dream. Thank you to all who are now a part of my world-wide Barth family.

(L-R) Florence Mannes, Susan Hone, Marc Sernel, Michaela Damin & Paola Cazzaniga
(Photo courtesy of John Wilkins [2016])

While at the conference this summer, we had a meeting of the boards of the Barth Syndrome Foundation and its four affiliates. This was the first time all the boards met together, and it was so nice to have everyone in one meeting sharing their ideas and accomplishments. Hopefully, we will be able to have more of these meetings at future conferences.

On the home front, we had a family activity in September along with our annual planning meeting. We will again be participating in Giving Tuesday, as we had great success raising funds on last year's day. Although we have no major fundraising event planned for the near future, we are hopeful that with personal fundraisers, the annual mail appeal and Giving Tuesday, we will be able to raise enough funds to continue funding research.

I would like to encourage families to periodically check our website and/or Facebook page for information on upcoming research/studies they can participate in. Without the participation of individuals with Barth syndrome and their families, this research cannot proceed to the next level. This is an exciting time for BSF and its affiliates, with clinical trials expected to begin sometime in 2017. With everyone's dedication, I am confident we will find a treatment within the next few years.

Barth Syndrome Foundation of Canada Planning Session 2017

(Back row) Susan Hone & Carol Wilks
(Front row) Lynn Elwood, Chris Hope,
& Lois Galbraith *(Photo courtesy of BSFCa [2016])*

The Barth Syndrome Foundation of Canada (BSFCa) planning weekend was held earlier this year and was combined with a family outreach. It made for a very busy but worthwhile three days. My family accompanied me on the trip this year to visit our families, biological and Barth. I left them for two days and headed to our favourite meeting place at Lake Kashabogg, Ontario. Attending were Lynn Elwood, Chris Hope, Susan Hone, Lois Galbraith, Carol Wilks and our chefs/dishwashers, Les Morris (also Board advisor) and Adam. Missing was Cathy Ritter who could not attend as she was busy being a devoted mother to her very ill daughter. We missed Cathy's expertise, knowledge and smiling face terribly.

Saturday morning was spent updating those who could not attend the Barth Syndrome Foundation (BSF) conference in the summer and reviewing our previous year's accomplishments. With the advancements being made in possible treatments for Barth syndrome, it is important that we keep our Barth families engaged and well informed on research they can choose to participate in. Families are encouraged to check the website and Facebook pages for updates. Of course, research requires money, and we discussed what possible fundraisers we could do in 2017.

A boat ride around the lake Saturday evening revitalized the group for supper. As always, we were well fed and pampered by Les, Lois, Adam and Carol. One again, they donated the groceries and accommodations to make our weekend cost free so that all our donations are used toward our mission of *Enhancing the lives and outcomes of Canadian individuals and families affected by Barth syndrome.*

A Day With My Barth Family

By Sheldon, Affected Individual (age 15), Ontario, Canada

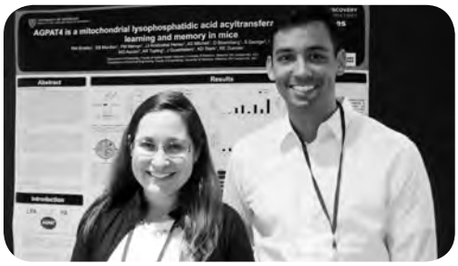
On September 17, 2016, I went to the annual get together (Outreach) for the Barth Syndrome Foundation of Canada. I have been going every year for the past six to seven years, and, as usual, it was a fun time. It all started with the visit to Ripley's Aquarium in downtown Toronto. Now, even though I live in Toronto, I never got the chance to go, and I thought it was different in a sense that there are things there that you don't see at other aquariums. There were a lot of different attractions like Dangerous Lagoon, and, of course, a big variety of aquatic animals. Also, to be honest, I don't think I would've been able to find the time to go if it wasn't for the get together. The aquarium was great, but we had to leave to go to The Old Spaghetti Factory.

I've never been to The Old Spaghetti Factory, although this time it's because it's in East Toronto. When we got to our table, we spoke with each other for a little bit and then got to order our food. After eating, we discussed some things going on in the Barth community and different drugs/treatments that could help us in the near future. Getting together with the group is always a great time. We talk, take pictures, and I liked seeing and meeting other people who have the same experience as me. All in all, it was a wonderful time, and I can almost guarantee that next year will be the same.

(L-R) Sheldon (age 15), Travis (age 21),
Adam (age 27), Jared (age 22) & Robert (age 30)
(Photo courtesy of BSFCa [2016])

Personal Perspectives of the Barth Syndrome Foundation Conference

By Robin Duncan, PhD, Assistant Professor, Department of Kinesiology, Faculty of Applied Sciences, University of Waterloo, Waterloo, Ontario, Canada



Dr. Robin Duncan & Ryan Bradley

In my scientific career, I have never experienced anything like the Barth Syndrome Foundation (BSF) conference this summer. It was an honour and a privilege to be invited, but it was truly a gift to attend. I will try my best to do justice to this wonderful experience.

I should first speak about the excellence of the SciMed sessions. It was two full days of "Barth 101." I got a rapid refresher on the molecular science, which is my focus, and I also got to speak with the small pool of international scientists working on cardiolipin synthesis, since everyone who has discovered a relevant enzyme was there. The face-to-face time was invaluable for comparing notes, approaches, and challenges. Even beyond the molecular work, though, the clinical SciMed sessions were better than I ever could have hoped for. These are competitive times in research, and I heard more than one person remark that this group works together, despite incentive to do otherwise. This was so true. The genuine interest, and open sharing of data by researchers was, in my experience, unprecedented.

Perhaps most importantly, however, by attending the conference, I got an incredible education in the personal course of this disease. As scientists, we read and read, and often find ourselves mired in the details. We see symptom as clues and can miss the individual cost. It was so inspiring to meet and hear from the talented, funny and caring young men experiencing Barth syndrome and to speak with their devoted friends and families. And as a mother of three young boys, it was also very personal. I've never cried at a conference before! In the end, I was left with a sense of deep honour and gratitude to all supporters of the Barth Syndrome Foundation, for entrusting my research program with the opportunity to hopefully make a small difference. I'll end by offering my warmest thanks to all members of the BSF and BSFCa for such kind support and for an experience that left me both optimistic and energized.

This was Dr. Duncan's first time attending the BSF International Scientific, Medical and Family Conference. Dr. Duncan was invited to present her work at the SciMed sessions. (Photo courtesy of BSFCa [2016])

Conference Reflections

By Ryan Bradley, PhD Student, Physiology & Nutrition, University of Waterloo, Waterloo, Ontario

(L-R) Susan Hone, Ryan Bradley & Lynn Elwood *(Photo courtesy of BSFCa [2016])*

As a scientific trainee in molecular biology, it can become very easy to forget that the research I am doing holds significance for families around the world. Often times, I find myself more focused on pathways and proteins than on the clinical outcomes that my work may help create. I was fortunate enough to attend the 8th biennial Barth Syndrome Foundation conference this past July, and it was truly an unforgettable experience. Due in large part to my fellow attendees, both scientists and patient-families, from the minute I first walked in, I felt right at home. The Barth Syndrome Foundation did a fantastic job of not only organizing Scientific and Family sessions but also by welcoming every attendee and making them feel part of something bigger. As much excitement as there was about the advances in research that had been made since the last meeting, the excitement everyone had in seeing one another was palpable.

I was able to present some work from my doctoral studies during the poster sessions and received great feedback and questions from scientists, patients, and their families. There were many unique insights and ideas that I am now implementing in my current research. The Scientific and Medical sessions are among the best series of talks I have had the opportunity to attend in my career so far.

I now enter the third year of my PhD recharged and refocused from the BSF conference. Since July, I have been proudly wearing the Barth Syndrome Foundation "Grow Stronger' bracelet, and I will continue to wear it until the end of my doctoral studies, at the very

(Cont'd on page 23)

Conference Reflections

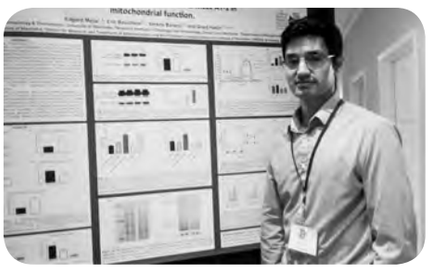
(Cont'd from page 22)

least. It is a great honour for me to be able to apply my knowledge and research abilities to try and make a difference not only in the lives of these young men but for their families as well. I look forward to seeing you all again in 2018!

Ryan Bradley presented two abstracts at the Barth Syndrome 8th International Scientific, Medical and Family Conference Poster Session.

A Return Experience

By Edgard Mejia, Post-Doctoral Fellow, Department of Immunology, University of Manitoba, Manitoba, Canada



Edgard Meija

As a second-time attendee of a Barth syndrome conference, I had the privilege to personally witness how much this meeting has grown in a short period of time. Not only were there more posters than the 2014 meeting, but there was also a greater variety of research. This year's meeting showcased some of the most promising Barth syndrome research I have ever seen. The work that was previously presented at the 2014 meeting has progressed tremendously. In the span of only two years, scientists have been able to produce an incredible amount of data that has contributed to our understanding of Barth syndrome. In addition, it was also very exciting to see the research being conducted by some of the newer investigators.

Attendees of this year's conference were able to see everything from improved methods to diagnose patients, to the use of advanced scientific techniques to discover new ways to treat this disorder. New discoveries are still being made, and this is representative of the hard work of many individuals. One of my favourite aspects of this conference is the fact that I get to interact with researchers and other students. Seeing how excited everyone is about their work is a source of motivation for anyone working in Barth syndrome research. This is augmented by the fact that students and principal investigators get to interact with the families of patients suffering from this disease. Talking with a number of families helped me understand that more work is still required and that this research is of utmost importance. I am confident that better treatment options will be developed soon because there are many people that are passionate about this work. As always, this was an amazing conference that was well organized, and I am glad I was a part of it.

Edgard Mejia has attended two BSF conferences and submitted posters each time. (Photo courtesy of BSFCa 2016)

My Perspective of the Conference

By Adam, Affected Individual (age 26), Ontario, Canada

Adam (age 26)

BSF conferences are always great, and this one was no exception. It is always fun to have a week with my mom, Lynn, to sit and catch up.

The session with Dr. Matt Toth, Science Director for BSF, with just us guys about therapies was great — so great that I went to it twice. Matt simplified a very complex subject. and I was happy to "see some light at the end of the tunnel."

Some of the older guys asked BJ Develle (a BSF Board member) if we could sit around and talk about real life issues that we face. We all know about and have discussed Barth syndrome a lot, but it was great to discuss situations and know that others experience and live with the same issues. BJ is always there for us. He is our "rock"!

Socializing at the conference is special. It is always great to see old friends again. We do not talk for two years, but we just pick up where we left off. I met young guys with Barth syndrome, and I know they do not know what is going on. Maybe with the new therapies, they will grow up without Barth syndrome.

The "Pill- Swallowing" Clinic has made a world of difference for me. Instead of enduring pain, I can now actually swallow medication — this is huge for me. I am also happy to say that I will be going to St. Louis to take part in Dr. Cade's study.

This year's conference was THE BEST!! *(Photo courtesy of BSFCa 2016)*

Association Barth France

Becoming an Advocate for Those with Barth Syndrome — A New Tool to Help Parents... and our Boys

By Florence Mannes, Chair, Association Barth France

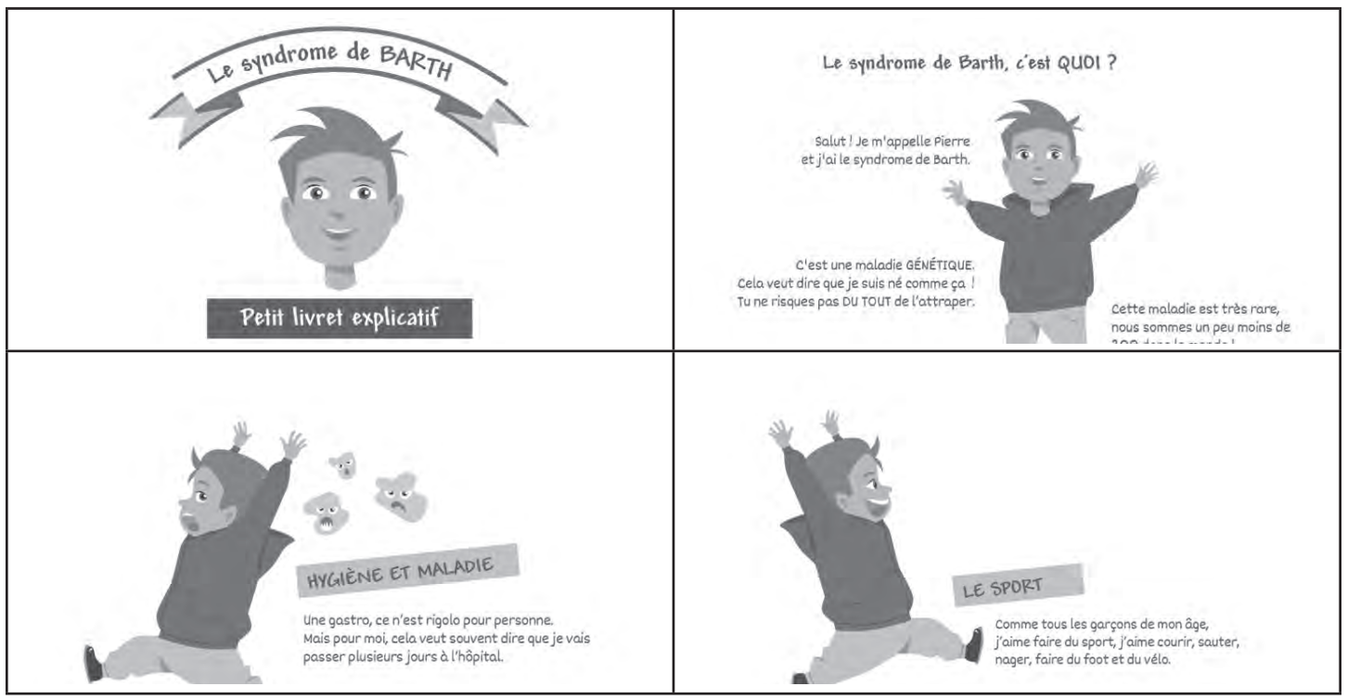
Raphaël (age 7) & Florence *(Photo courtesy of Amanda Clark [2016])*

Explaining Barth syndrome is always a bit tricky: you don't want to be too dramatic, but on the other hand, some people who are living with our boys need to know, exactly, what Barth syndrome means.

When you talk to children, the balance has to be even more precise: you don't want to frighten them, but they need to understand what is going on with their friend.

Every year, when the school begins, we, as mums of boys with Barth syndrome, have to become advocates for our sons to their teachers, school directors, and friends. I've been doing that since my son first went to school, five years ago; but last year, I felt we were missing a tool that could help us. So, in order to prepare for this new school year's introduction to Barth syndrome, we decided to create a small book talking about Barth syndrome in an easy and fun way. Our very first idea was to provide schoolmates of our boys with a little book on Barth syndrome they could understand easily. We wrote the text carefully, trying not to forget any aspects of the syndrome, but trying to avoid being too dramatic. We then asked Zoé, a close friend who is an illustrator, to provide us with the drawings that would match the text…

This is what we ended up with (these are a few pages taken from the entire booklet)!



300 copies of this booklet have been printed, and it is also available online on Barth France's website. It has been sent to all the French speaking families (in France but also in Belgium and Quebec) who have children below the age of 12. Families have found them very useful when talking about Barth syndrome, not only to peers of the boys, but also to the adults who are dealing with our boys every day. The booklet usually remains in the classroom for the school year; the kids are happy to discover it at their own pace.

We had some discussions with the other BSF affiliates and hope this booklet will be translated into English and Italian. Our next step is to prepare the same kind of tool for teenager Barth boys to help them become advocates for themselves.

On a personal note, I wanted to share the discussion I had mid-September with my seven year old boy with Barth syndrome. One night,

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Becoming an Advocate for Those with Barth Syndrome — A New Tool to Help Parents... and our Boys

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he came back home from school a bit sad and concerned. He finally explained to me that a new girl in his class made fun of him because he was so small and not able to write as other kids do. After explaining to him that he needs to be stronger than that and that he should try to focus on what his friends are saying instead of on the ones who are bullying him, he still did not seem satisfied with my answer (which, I admit, was far from perfect…I was not yet ready for this type of concern). Half an hour later, he came back to me, saying that he wanted to present Barth syndrome in front of his class "to make sure the girl understands exactly what Barth syndrome means"… and that's what he did, with the support of the printed version of the booklet. Since then, he hasn't complained about peers laughing at him… I hope this will work for the whole year!...

Raphaël presenting Barth syndrome to his class

Activities of Barth France: The same events, each year bigger

Organizing fundraising events is very time consuming…and when, as an Association, you have some events that are successful, you do your best to improve those, before trying new ideas. That's how we ended up with organizing the **6th Barth France Golf Tournament** in August 2016, the **5th Edition of the Barth France Poker Tournament** in November 2016, and the **3rd Edition of the Black Truffle Gala Dinner** in January 2017. The Golf Tournament gathered 90 golfers and allowed Barth France to raise up to Eur 10.000. Around 140 people are expected at the Poker Tournament, and we have to limit to 200 the number of participants to the Black Truffle Gala Dinner in January to make sure the quality of this event complies with the donors' expectations. *(Photos courtesy of Barth France [2016])*

6th Barth France Golf Tournament

5th Edition of the Barth France Poker Tournament

Spreading the word, meeting new people, meeting brilliant minds

Life is a matter of encounters, and that's what makes it so lively. You never know what to expect from the next day….I have at the back of my car a Barth Syndrome Foundation sticker…which, one day, about one year ago was noticed by someone in the street. This man first made a donation to Barth France, which was great! …but that was just the beginning of a much more powerful partnership. He works for the Atomic Energy Commission (CEA) in France, more precisely for a branch of the CEA called "Institute of Research into the Fundamental Laws of the Universe"…which seems to be really far from Barth syndrome…CEA is one of the key players in research, development and innovation in four main areas, among which is *"fundamental research in the physical sciences and life sciences."*

As the CEA regularly organizes conferences on wide and various subjects for its researchers, a conference on Barth syndrome was finally held in September at the CEA, thanks to the motivation and willingness of just one man, met through a sticker at the back of my car…

Four different aspects of Barth syndrome were presented during this meeting:

- Cardiomyopathy in Barth syndrome (by Damien Bonnet, head of Pediatric Cardiology at Necker Hospital for Sick Children)
- Hematology aspects of Barth syndrome (by Jean Donadieu, pediatric hematologist at Trousseau Hospital)

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Becoming an Advocate for Those with Barth Syndrome — A New Tool to Help Parents... and our Boys

(Cont'd from page 25)

- Potential roles of genomic medicine in rare genetic diseases such as Barth syndrome (by Jean-François Deleuze, head of the French Centre National de Genotypage)
- Everyday life with Barth syndrome (by Florence Mannes, mother of a seven year old Barth boy).

(L-R) Damien Bonnet & Jean Donadieu

Jean-François Deleuze

These presentations led to multiple questions and discussions between doctors and scientists and should lead to new perspectives in research on Barth syndrome. *(Photos courtesy of Barth France [2016])*

Barth Italia
Attending the Conference was Therapeutic

By Paola Cazzaniga, President, Association Barth Italia

(L-R) Pietro (age 15), Paola & Paolo
(Photo courtesy of Amanda Clark [2016])

This was the first time we'd taken part in the Barth Syndrome Foundation conference both as a family and as a founding member of Barth Italy, and it was a really intense experience from every point of view.

When we came back, I was pleased to be able to share what it meant personally and as a family with the other families of Barth Italy. It helped give a positive focus to our future initiatives, how we would inform schools and hospitals. It motivated us to create new projects with our families and gave us a further push in our fundraising efforts. All this has been made clearer by the hope and the powerful affection we got from attending the conference.

I have to say that, before attending, each one of us had our own fears.

My son, Pietro, was worried that he wouldn't be able to face up to the problems of language, and in general he was worried he'd feel excluded from the group of other Barth boys who know each other well. He also was worried that we would be disappointed if we saw that he wasn't able to enjoy the experience. At 15 years old, he knew how demanding, in all terms, this trip was for us.

My husband, Paolo, was very worried by the idea of meeting the older boys, seeing firsthand the possible painful future for our son, and by the idea of having to share with other families the suffering which for years Paolo has handled with difficulties in his own private way. Paolo felt he might be hurt by being part of the world of explicit suffering from which he has defended himself and which has often prevented him from sharing his feelings with me even though he has always, always, always been by my side.

I was worried that I would lose myself in a kind of euphoria and my drive to do things which often occurs and helps me to face a feeling of inadequacy in certain situations. I was also afraid I might find myself in the situation where positive emotions and their collective encouragement wouldn't leave space for my husband's pain and my own tears. It's incredible how we project our fears and worries into our future outlook.

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Barth Italia

Attending the Conference was Therapeutic

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Our arrival at the Hilton Hotel in Clearwater Beach, Florida made us forget how long and tiring our journey had been. There was a beautiful white beach of fine sand, a sugar beach, with palm trees, sun and sea.

(L-R) Carmela Lauretti, Paola Cazzaniga & Anna Dunn *(Photo courtesy of Paola Cazzaniga 2016)*

We met Shelley Bowen (Director, BSF Family Services & Awareness) for the first time, and she was in really busy mode. She welcomed us affectionately with her husband, Michael, and gave Pietro a bracelet so that the others would recognize him. She introduced us to several families around the swimming pool. Shelley is so helpful for moms visiting for the first time. She has patience even about explaining the progress being made in understanding the illness and its treatment. This was all so incredible for me.

The other families were equally wonderful. It wasn't difficult for us, despite cultural and language differences, to feel part of Team Barth straight away, proof that affection can move huge hurdles.

Pietro was quite shy and didn't know a lot of English but jumped into the pool with the ball making friends with the Barth boys, brothers and sisters. They welcomed him into a lovely group of adolescents, and they helped him with his fears. He took part in the clinical tests and had a great time. He just kept going for an entire week, except when he went back to the room to sleep or to get more dollars to order food not knowing what he was going to get!!

Paolo and I got to meet wonderful people who crossed the United States with their whole family to take part in the conference. Others came from Australia, Canada, France and England, all there to meet the others and work hard together for the week, during which everyone was able to offer their personal contribution.

The Welcome Ceremony was especially helpful for first-time families like ours. The whole week was really affectionate and warm and gave everybody a chance to get to know each other, to tell our story, to exchange feelings, and make suggestions to the Foundation, as well as offer advice to our kids. But it was also a chance to have fun, laugh together over meals and be worry-free for a while.

A special thank you goes to Anna and Mark Dunn, who have Italian roots. They, together with their son, Aldo, and his Grandma, Carmela Lauretti, were our guides, taking us around and looking after us wonderfully during the whole experience which was intense and at times trying and moving.

BSF's Science Director, Dr. Matt Toth, was also incredibly approachable, and he illustrated with the help of Anna the most recent research by BSF and its future prospects. It's wonderful to understand that there is real hope!!

The strongly compassionate atmosphere which was created got rid of my fears and let us feel free to express ourselves as best we could; when we couldn't find the words, hugs and looks were enough.

It was also wonderful to see how the Italian scientists, Dr. Morrone from Florence and Dr. Corcelli from Bari, took part with drive and verve.

I often felt fear and vitality, pain and happiness, but, for the first time, I didn't feel bad about crying and then feeling I wanted to dance. My husband also wept with a friend, not knowing what our future will be, but also understood this agony won't kill us. There is hope, and sharing that helps.

Last but not least, Pietro, who was worried about feeling alone, thanks to the friendships made with the boys, understood that he can do things even on his own, if he puts his mind to it.

As a therapist, I asked myself what determines change in a person? What factors allow one to re-organize inside, not before and not after, but in that precise moment in life when it makes sense to change? I have seen professionally that the work of months and years can be brought together by a specific experience: it's like having all the pieces of a jigsaw form a complete picture at one precise moment. I can honestly say that, for us, the conference was the kind of therapeutic event which allowed us to share who we are and who we want to be.

My experience as a chronically unsure Barth mom has often led me to a vortex of worry, insecurity, and impotency in which I ask myself if I'm doing everything possible, if it's enough to look after my son as I do. Well, among other moms like me, I think I can say that I'm doing everything I can. I'm doing my best, and I feel at peace with myself.

I have recounted all this to my Italian families. I've said that when you go through experiences like those of our families, it is difficult to make sense of it all, but it is still important that we believe, as part of Team Barth, that we're on the right road. I really believe that we can start to make sense of our pain, helping those who feel alone and overwhelmed, and make our small contribution to research, together with Barth Italia. Thanks to all of you!

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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)*

Deacon (age 1)

Kevin (age 27)
(Photos courtesy of Amanda Clark [2016])