Barth Syndrome Foundation of Canada

Canadian Newsletter

www.barthsyndrome.ca

November 2018

Inside this issue:

President's Report—Fifteen years of devotion and commitment *by Susan Hone*

It is that time again when we take a moment to sit back and reflect on events and proceedings throughout the past year -- what worked, what could be improved upon, and where to go from here.

Susan Hone and Dr. Colin Steward (England). *Photo by Chris Hone*

This November 20th we celebrate Barth Syndrome Foundation of Canada's (BSFCa) 15th Anniversary. This is quite an achievement. While it is relatively easy to start a foundation, keeping it going requires people with passion, persistence and dedication. So many people have given of themselves to help us reach this point. Everyone involved with this organization is a volunteer, and without them we would be lost. We truly appreciate not only financial donors, but also donations of time.

The biggest event this year, of course, was the 2018 Scientific, Medical and Family Conference, both for individuals, families, and the Foundation. While members of Barth Syndrome Foundation of Canada support the conference individually as volunteers or sponsors, we also contribute as a proud affiliate. It is one of the most important programs that we invest in financially. We sponsored the Photo Booth at the Friday night social which is always a big hit with families, the Poster Session (during which researchers interact with each other and subsequently with families to discuss their investigational outcomes and future ideas), and a luncheon for both the families and professional attendees. In addition to the sponsorship contributions, we also provided flight and hotel accommodations for two Canadian researchers to attend.

The conference provides families with valuable information from other conference attendees, doctors and scientists. It is a one stop shop on Barth syndrome. Over seven days, the conference includes new family orientations, a Barth essential information session, sessions on neutropenia, cardiac aspects of Barth syndrome, genetics, carrier support, techniques for daily living by age groups, research clinics to

Individuals affected by Barth at the 2018 conference. *Photo by Amanda Clark*

(Continued on page 12)

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

9th International Scientific, Medical & Family Conference *by Chris Hope*

This past July the Barth Syndrome Foundation (BSF) hosted its ninth conference. It was at the first one in 2002 that the idea of creating a Canadian Barth foundation was first discussed.

Much has happened in the intervening time—we are now one of four affiliates around the world (UK, France, and Italy) all working towards the same goal—finding a cure for Barth syndrome, which all too often remains a fatal disorder.

While affiliates run their own agendas, we all come together on two of BSF's major programs. One is to contribute financially where and when we can towards research grants that have been approved by their Scientific and Medical Advisory Board, and the other program we help support, as much as possible, is the biennial international conference.

Each conference gets bigger and better, and, of course, this last one was the biggest and best yet. This year over two hundred and twenty-three family members traveled to Florida from twelve nations.

The conference started with the traditional research and individual appointments, during which time families and individuals with Barth syndrome were able to ask questions and get advice from the true experts of the disorder. In these two and a half days, over 500 research and consultation appointments took place, in between, small group sessions in which families were able to learn and share experiences.

On Thursday and Friday formal lectures were held covering all major symptoms of Barth syndrome. We are fortunate to have so many renowned doctors and professionals give up their time to come to the conference to inform, clarify and even learn.

After a full day of lectures, Drs. Barry Byrne and Christina Pacak graciously responded to families' requests, and gave more of their time to explain gene therapy in layman's terms. It is still incredible that this may be a reality for Barth syndrome, and that we will hopefully be looking a starting a clinical trial within the next few years.

On Thursday, the poster session, which BSFCa is proud to sponsor, was again a popular event for both professionals and families. This was again followed by the moving 'luminaries on the beach' tribute.

Once again, the line between medical professionals and patients became a little blurry during the traditional Friday night social. Everybody enjoyed the dancing and entertainment, while the photobooth, which BSFCa sponsored, was once again a hit, and was occupied with groups of merrymakers from start to finish.

Canadians and a few wanna-be Canadians at the conference. *Photo by Susan Hone*

After a full week of almost constant activity, it was time for the closing ceremony. The conference was ended with a moving video of photographs taken throughout the week, and with testimonials from families and individuals about the impact the conference has made on them.

History in the Making *by Chris Hope*

PFDD Panelists. *Photo by Susan Hone*

On Wednesday, July 18, 2018, BSF made history. Of the more than 7,000 rare diseases, BSF became the 14th organization to host an externally-led patient-focused drug development meeting (PFDD) with the US Food and Drug Administration (FDA). The purpose of this meeting was for affected individuals and their families to create the Patient Voice for Barth syndrome for drug developers. This gave BSF the opportunity to inform the decision makers what it is really like to have Barth syndrome, or to care for someone who does. Following FDA procedures, panelists and speakers (affected individuals and caregivers) spoke about their symptoms and about their difficulties in coping with daily living, and they gave their viewpoints and hopes of current and future treatments.

Families and individuals frequently get into the habit of coping with Barth syndrome step by step and try to downplay or cover up the difficulties they experience. These panelists were honest and spoke candidly about their trials and concerns. In preparing for this meeting, they had to face their own fears and uncertainties, and all spoke eloquently and powerfully in their allotted time.

The panelists were chosen to represent several different stages of development of the affected individual, covering infant, preschool, elementary and high school, college, post college/work, and older. They were asked to talk about their greatest anxieties and which areas of concern they most wish to be addressed by doctors and researchers. While the expected fear of an early death and wish for a healthy heart were mentioned, surprisingly most panelists stressed the quality of life issues, specifically overwhelming fatigue and muscle weakness were priorities.

For many in the audience, one of the most difficult ten minutes of the afternoon, was a video dubbed "Uncle Bob". Bob is an individual with Barth syndrome in his fifties. He lived most of his life as an active farmer. Several years ago, he needed to quit working, and his muscle weakness has since deteriorated quickly, and he is now in a full care facility, fully dependent for his daily care. While this is the story of only one man, it is a call to action to accelerate research and treatment plans since the future of Barth syndrome is still a mystery.


Robert asking a question at PFDD meeting. *Photo by Susan Hone*

This five-hour meeting could be one of the most important that has ever taken place to advance and increase doctors' understanding of Barth syndrome, and what patients really experience and need. Many doctors commented that although they have been coming to the conferences for years, and have

(Continued on page 5)

Caleb's First Conference *By Jasmine, Mother of Affected Individual*

Caleb. *Photo by Mom Jasmine*

In his first six months of life, Caleb suffered cardiac arrest, he was put on ECMO as his heart stopped beating for five days, he was on life support for two weeks, he was put in isolation due to a common cold virus, he received numerous doses of medications that you'd never imagine giving a child, he was poked and prodded on a daily basis for several months straight, he was intubated for 93 days because he couldn't breath on his own, his chest and head got so swollen that the fear of a brain aneurism was very real. He wasn't expected to live.

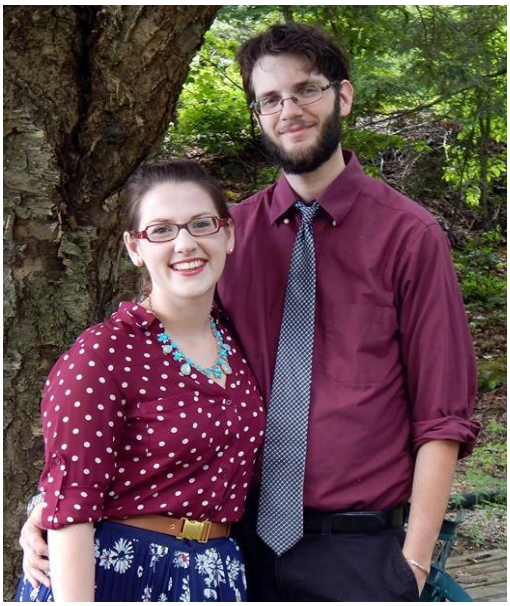
Part way through those first six months, I chatted with an amazing lady who worked within the ICU. I told her how I had found the Barth Syndrome Foundation and explained to her how we'd connected with some families and found out about a medical conference that is held every two years. The conference is an opportunity for doctors, specialists and families to come together to inform, bring awareness, and fight for a cure. This conversation was at a time when Caleb had started to make some progress but still had a long way to go. She said to me that she had a feeling we would be sitting at that conference in 2018. I'll be honest, at the time, I wasn't so sure. I didn't quite believe it.

Fast forward a year, and we have just returned home from the 2018 Barth Syndrome International Conference! Ten days ago, we prepared to fly from our home on Vancouver Island to Clearwater Beach, Florida. I was a nervous wreck. Completely anxious and nervous to meet the families that I'd been chatting with on Facebook. We joined approximately 300 people (including 50 affected Barth individuals) from around the world to connect, to share our stories, to participate in trials and studies, to fight for a cure, and to build HOPE. Over the six days of the conference, we had the chance to listen to so many specialists and researchers and learn more about this rare disease. We learned about drug trials and gene therapy, we had lunch with brilliant doctors, we drank wine on the beach with top doctors from other countries, and we danced to "We are family," with researchers from around the world. We heard from other families struggling with this disease and learned that there are others living with the same burdens of Barth syndrome as we are.

In regards to the medical side, the conference was invaluable for us. We made some amazing connections, learned more about Caleb's syndrome, and were able to ask questions and get answers from people who focus specifically on this disease. Even more profound though, were the connections with our Barth family. From the first moment we walked down the hotel hallway, we were literally welcomed with open arms. Everyone knew exactly what we were going through and was willing to share and relate with their own stories and experiences. The isolation that we've felt in regards to Caleb's diagnosis was somehow lifted, and we knew we were with our extended family. From birthday parties on the beach, to late night swims in the pool, group selfies, endless tears and emotions, luncheons and photo booths; being a part of this group was an amazing feeling.

(Continued on page 8)

Life *by Ryan Ritter*



Jessica and Ryan. *Photo by Ryan Ritter*

I always find it kind of ironic and funny when people ask me what it's like living with Barth syndrome. Whether it was as a kid or now as an adult living with my girlfriend, Jess, and working full time. I don't have anything personal to compare it to since I've had it my whole life. I've never known anything different. It's like asking someone to compare two different meals. One they have had all their life and quite enjoy versus one they can only see. It's always seemed kind of a fruitless endeavour to me. "Ah look how much better that apparently is; just because I can't have it and it's not just like mine." It makes no sense.

When Susan first asked me to write this I had no idea what to say. So, I spent a week watching the people around me and talking to them about their experiences. I saw people worrying and stressing to make ends meet, trying to make it through the day, but having fun and goofing around with their friends and co-workers while doing it. Wishing they could spend more time with family. In short, I watched them do and experience all the things I do on a day to day basis. Yes, there are things I'll never be able to do and that is something I have come to accept from an early age. I would say, however, that that is true of just about everyone.

There are many things I can still do. I went to college, I can live on my own and pay my own bills. I hang out with friends and work full time. In short, I would say that, in my experience, there is no difference between living with Barth syndrome and living without it. True, there are more concerns and risks. But there are in everything. That's why it's called life and not hiding. I feel like the more appropriate question would be: "How do I let Barth affect me and my life?" And the truth is… I decide not to let it. I do everything I can and want to. There are days when I get home from work and am exhausted and days I feel like garbage. There's no avoiding it. What I have to keep in mind is that tomorrow is a new day and I need to make the most of it. If it ends up being the last thing I do… then screw it. I refuse to let Barth hold me back from the life I want to live, I will not regret the chances I didn't take.

History in the Making *by Chris Hope*

(Continued from page 3)

treated patients with Barth syndrome, this meeting was a real eye-opener, and they came away with a lot more knowledge and information than ever before. The fact that living is just as important as quality of life has given researchers the impetus to re-examine their approach and expected outcomes of their research.

We are now waiting for the *Voice of the Patient* report, which is scheduled for publication in a few months, and will be on the FDA website. This report will be used to inform researchers and other interested parties to prioritize clinical outcomes and to fast track approvals for therapies.

From the Heart *by Les Morris and Lois Galbraith*

This article is from the hearts of all of us who were privileged to know Susan McJannett!

The men in Susan Marion McJannett's life, husband Bob and sons Andy and Rob, described the marvelous woman beautifully in a tribute:

Susan and Marty. *Photo by the McJannett Family*

Entrepreneur

Automobile Enthusiast

Volunteer

World Traveller

Advice Giver

Red Hat Society Member

Adequate Choir Singer and a

Pretty Good Equestrian

Susan. *Photo by the McJannett Family*

They also said "Susan McJannett reached the end of her mortal road on Wednesday May 9th, 2018. We like to think she is now cruising in her 1958 Pontiac Parisienne, two-door hardtop."

The BSFCa can add many more adjectives - ***Volunteer Extraordinaire*** comes to mind immediately. She and Bob were the dynamic duo that were forever coming up with new and creative ways to really have some fun and raise a few bucks at the same time for the BSFCa. There were countless dances because Susan and Bob loved to listen and dance to great Canadian bands. They arranged for silent auctions and raffles and always found matching funds for all these endeavours. They always rallied their automobile club friends and the Buss Megg Society.

At annual golf tournaments Susan volunteered at the registration desk, sold mulligans and golf balls and greeted each and every golfer with a warm smile. Along with her friend Carol, Susan then spent the day on the course as the 'Official Golf Foursome Photographer'. Again she took the opportunity to chat up the golfers and spread the warmth of her Barth spirit!

Susan was an honest and 'real' person and we are thankful for her love and support of the BSF of Canada. Her enthusiasm and exuberance and love of Barth still lives in all of us.

Volunteer Extraordinaire - Friend Extraordinaire!!

Barth Syndrome Foundation Saved my Son's Life *by Lynn Elwood*

Adam and Les. *Photo by Lois Galbraith*

You just never know when a crisis will occur and you may need help. The good news is that whenever you do face that situation – the Barth Syndrome Foundation (BSF), its affiliates and the families are there to help.

In late April last year, I got one of those phone calls you hope never to get. My Father called to tell me my son, Adam, was in a serious ATV accident. He would be okay but was "banged up pretty badly" and was being airlifted from the local hospital near our cottage to the best trauma hospital in Toronto.

We sprang out of bed and into action. First, I grabbed my Barth go bag – which has been packed and by the bedroom door for many years (thank you BSF!). Next, I picked up the huge binder of Adam's medical records (thank you Kate McCurdy and Shelley Bowen who impressed upon me the need to have this together). Next, I went to the website and printed off several of the FACT sheets. Then we headed off to the hospital to wait for the helicopter.

As we waited for Adam to arrive, we shared a pamphlet and some information sheets with the trauma physician on call. They asked some questions and took it all very seriously as they prepared for his care upon arrival. This was the start of sharing information from BSF with dozens of physicians and healthcare professionals as they worked on Adam. We were extremely impressed by the level of attention they paid to the material and to our guidance – and we believe it saved Adam's life.

Once the initial assessment was done by the trauma team in emergency, it became clear that Adam had fractured his C1 (top of the spine) and broken his jaw and several bones in his face. Serious injuries indeed and at least one surgery required. Fortunately, his neck fracture did not require a permanent rod in his neck, nor did it affect his mobility or neurological function. He was very lucky. Instead he needed to have a "halo", essentially a cage with pins into the skull and a stability vest that held his head and neck stable for several weeks until the neck fracture could heal. Adam did need surgery to put plates in his face and wire his jaw shut until it healed, then a minor procedure to remove the jaw wires.

From the very first, the Barth Syndrome Foundation, the parents and clinicians were there helping in so many ways. Susan Hone and Cathy Ritter gave me advice and the words I needed to convince the doctors and nurses to ensure he had glucose in his IV and especially before surgery. Shelley gave me contacts and advice, and Michaela put me in touch with Dr. Steward who provided the much needed guidance to the hematologists so they would be proactive with neupogen and keep it in his system until the halo was removed and the pin sites healed. Nicole Clayton, dietician in the UK, sent information on Adam's required daily nutrition amount, which allowed them to remove the feeding tube when his jaw was wired shut. Whenever there was something we needed to advocate for, there was someone or a written article to back us up so we could get the best possible care for Adam.

Of course, it was a tough time for Adam, but he was an inspiration. He treated people with kindness and humour, even when he was suffering. The hospital staff told me on more than one occasion that they

(Continued on page 9)

Abbie's Lemonade Stand! *by Cathy Ritter*

Abbie and Noah *Photo by Mom Stephanie*

When you are 5 years old and your mom brings home a super-duper craft to do… you get excited! And such was the case for 5 year old Abbigale (niece of Ryan, Bths) when THE Lemonade Stand entered their house. Only one problem. It was still early spring in snowy Central Ontario. No matter – more time to get prepared! Over the next few weeks the project was constructed, coloured, decorated, and a date was set.

On a sunny, windy summer day The Lemonade for Barth Stand was set up at the end of the driveway! Not only did it contain lemonade, but delicious, home baked and decorated cupcakes. (And for those who know Abbie's mom's prowess at baking… delicious, extravagant, over-the-top treats to be sure!). Over the next couple of hours neighbours, grandparents, and neighborhood kids came to buy some lemonade and a cupcake. All proceeds were donated to Barth Syndrome Foundation of Canada.

It was a lovely day – awareness for Barth syndrome was raised and young Abbie learned a life lesson in giving to others – not to mention the satisfied tummies from the lemonade and cupcakes!

Caleb's First Conference *By Jasmine, Mother of Affected Individual*

(Continued from page 4)

I'd like to thank Help Fill a Dream for sending us to this conference. Without them and their donors, we may not have had the chance to meet our new family. I'd like to thank all the people that worked so hard putting this conference together. I'd like to thank all the doctors and specialists who travelled to the conference not only to give speeches, but to mingle with the families and meet one on one to go over medical data. I'd like to thank those doctors and researchers who are working tirelessly to find a treatment or a cure for this dreadful syndrome. I'd like to thank all the families who shared their stories both joyful and painful. I'd like to thank those families for sharing their experiences of the everyday effects of having a child with Barth syndrome. I'd like to thank the other moms for the hugs as we discussed the guilt that we carry knowing we're the carriers of this disease. I'd like to thank the older Barth individuals for taking both of my boys in as your own little brothers and also for sharing their struggles with us despite how painful they were. These boys and men are the reason we left this conference with so much hope. They continue to fight for not only themselves but the young boys like Caleb and any future boys that find themselves living with Barth syndrome.


Caleb and family 2018. *Photo by Mom Jasmine*

To our Barth family, we are so grateful to have shared such an amazing week with you all. We were greeted with so much love, and we look forward to seeing everyone again in 2020!

Lighting up the Night *by Susan Hone*

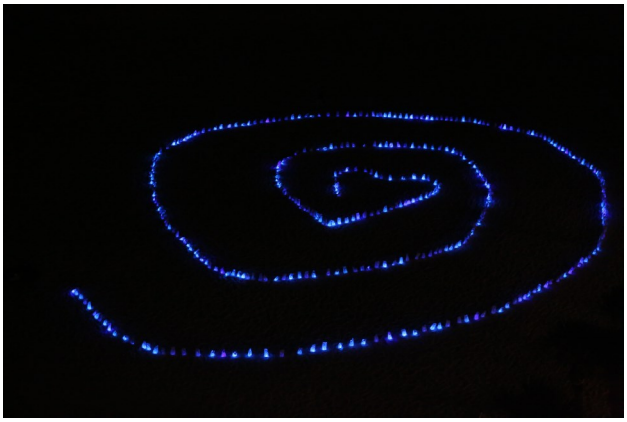
Overhead Drone Shot. *Photo by Dave Perry*

For the past few conferences mine and my daughter's family have sponsored the luminaries on the beach. Each luminary represents and honours the memory of individuals with Barth syndrome known to us. The luminaries are placed on the beach in the form of the Barth Syndrome logo and people are marched into the logo led by a bagpiper.

It is a very moving ceremony with lots of tears, hugs and smiles. This year, an added feature to the ceremony was a drone that flew overhead and recorded the event. The Grzesiak family graciously sponsored the drone. If you would like to watch the video of the ceremony , the link can be found by searching for "Dave Perry Barth syndrome" within YouTube or at:

https://vimeo.com/287122222

Piping in the Families. *Photo by Susan Hone*



Luminaries night shot. *Photo by Amanda Clark*

Barth Syndrome Foundation Saved my Son's Life *by Lynn Elwood*

(Continued from page 7)

enjoyed working with him. They invited him to speak with high school students that tour the hospital, and he did that several times, including his grandpa and mother, and impressed us all. We had more quality time together than normal over those months and there were some moments we will cherish.

Throughout the weeks as we nursed Adam back to health, our extended family were active partners – visiting often, ensuring he was getting calories (he even gained a few pounds while his jaw was wired shut), advocating with all the practitioners, bringing us laughter and sharing hugs. And our Barth family was there – only an email or a phone call away whenever we needed them - and understanding when we were too exhausted or consumed to be in touch. We are so very lucky to have such a wonderful family – those related by blood and marriage, and those we have gained through BSF and affiliates. Thanks to all of you and our close friends, my wonderful son is well again. We thank you with all of our hearts!

Clinical Trials, The Future is Now *by Chris Hope*

There are many stages on the road to finding a cure/treatment for Barth syndrome. One major step is a clinical trial. This is when a drug or a therapy is tested on a few human volunteers to determine the safety and effectiveness of the possible new treatment. These studies have strict guidelines on who can participate, and what the study involves. Everyone is checked at the beginning of the study, and is monitored throughout. All new treatments must be tested this way before they can be approved. It is a long trip just to get to the clinical trial stage. Only about one in 20,000 drugs that are developed through research get approved.

The current clinical trials that are either on-going, or are planned to start in the near future are:

Elamipretide—to improve mitochondrial function (underway)

Bezafibrate—to ameliorate the cardiolipin ratio (to start later this year)

Gene Therapy—to repair the underlying genetic defect

While reaching this stage is an immense achievement, it is only with volunteer participants that we will be able to keep moving forward. Benefits to volunteering can include gaining access to new research treatments before they become widely available, often at little or no cost, receive expert medical care at leading health care facilities (normally more frequently and thoroughly than usually), and you can help advance medical research and possibly improve medical care for yourself and others.

Dr. Grant Hatch, University of Manitoba. *Photo by Susan Hone*

Conference 2018

A few first-time conference attendees. *Photo by Amanda Clark*

Jared Hone's wheelchair gets a makeover. *Photo by Susan Hone*

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Adam Elwood and Iyar Mazar at the Poster Session sponsored by BSFCa. *Photo by Susan Hone*

President's Report—Fifteen years of devotion and commitment *by Susan Hone*

(Continued from page 1)

collect data from those with Barth syndrome as well as control subjects (brothers, physicians and scientists' sons) and scientists and physicians meeting to discuss their findings and compare notes along with various other meetings. My family has been fortunate enough to attend many of these conferences and the friends we have met have become our extended family. It is truly an opportunity every family living with someone affected by Barth syndrome should strive to attend.

Ryan takes over the photo booth. *Photo by Mom Stacey*

The other major contribution BSFCa makes is toward the annual grant process sponsored by Barth Syndrome Foundation Inc. In 2017 we contributed $25,000.00 US Dollars (USD) towards Christina Pacak, PhD, Assistant Professor University of Florida, Gainesville, Florida's study entitled "Optimization of AAV-mediated gene therapy for Barth syndrome". This year we allotted $10,000 USD to award Richard Epand, PhD, Professor Biochemistry and Biomedical Sciences, McMaster University, Hamilton, ON on "the cause and consequences of plasmalogen depletion in Barth syndrome".

On the home front, we have started to go through some changes in our board. In June, at the annual general meeting, one of our founding members, Christiane Hope, elected to step down from the Board of Directors. She remains heavily involved in the Foundation and is completely committed and dedicated to our mission and goal. Thank you, Chris, for your long-term service on the Board.

While board turnover and changes are daunting, the BSFCa is a strong organization, and we know that these changes can be rewarding. In order to not just survive but to flourish, it is important for this affiliate to anticipate change and look to adapt and tackle all challenges head on. We hope to recruit a talented new board member (or two) and with their ideas and dedication we feel that we will be able to move forward and increase our efforts in all areas.

For online donations, please visit www.canadahelps.org/en/dn/3811

What is Barth Syndrome?

Barth syndrome is a rare, genetic disorder primarily affecting males around the world. It is caused by a mutation in the *tafazzi*n gene which results in an inborn error of lipid metabolism. This error causes, in various combinations and varying degrees: cardiomyopathy (disease of the heart muscle), neutropenia (an abnormally low count of a type of white blood cell that helps fight off infections), underdeveloped skeletal musculature and muscle weakness, and severe growth delay and exercise intolerance.

While much progress has been made in treating Barth syndrome, unfortunately, it still remains all too often a fatal disorder.

Going Green!

If you have any concerns or are interested in receiving our newsletter via email, please contact us at info@barthsyndrome.ca