



Barth Syndrome  
Foundation of Canada

# Canadian Newsletter



[www.barthsyndrome.ca](http://www.barthsyndrome.ca)

March 2020

## Inside this issue:

President's Report	1,12
A Barth Bear's Adventure	2
An Interview with Travis	3,8,10
Virtual 5K Run Fundraiser	4,5
2019 Finance Report	5
2019 Outreach	6
2020 International Conference	7
Research Update	9
GE3LS HSPR Conference 2019	9
Friends, Cars and a Spectacular Birthday	10
In Appreciation	11
Glass Turkey Fundraiser	11
Online Donations	12
What is Barth Syndrome?	12
Going Green	12

## President's Report *by Susan Hone*

Welcome to 2020! Another year has passed by very quickly and I would like to update you on some of the Barth Syndrome Foundation of Canada's (BSFCa) accomplishments in 2019.

One of our main goals is to be able to contribute funds and participate in research being done into causes and potential treatment for Barth syndrome. We have been very fortunate to play a part in the Barth Syndrome Research Grant Program thanks to our loyal donors. The grant recipient we helped partially fund (along with Barth France and Barth Syndrome Foundation, Inc.) in 2019 was

Christopher Y. Park, MD, PhD, Associate Professor, New York University School of Medicine, who is researching the Characterization of hematopoietic stem and progenitor cells in Barth syndrome. BSFCa's contribution towards this \$100,000 USD grant was \$20,000 USD.

The 2020 grant cycle is currently underway. BSFCa will review the recommendations of the Science and Medical Advisory Board of the Barth Syndrome Foundation, Inc. to decide which grant we would like to participate in this coming year. We have allocated \$20,000 USD towards the grant cycle for this year.

Another major goal of BSFCa is to support Canadian families affected by Barth syndrome. To that end, an Outreach was held in May, 2019 to bring individuals, families and friends together for a weekend. This was our biggest Outreach to date. See



John and Jared at 2019 Outreach.  
*Photo by Susan Hone*

page 6 for more information. We are planning on another fun day this fall, and will be sending out information as plans are made.

We are excited to announce a new member to our Executive Committee, Jasmine Champagne. Jasmine lives with her husband, Mark, her three-year-old son Caleb who is affected by Barth syndrome, five-year-old son Jordan and two exchange students in B.C. Welcome

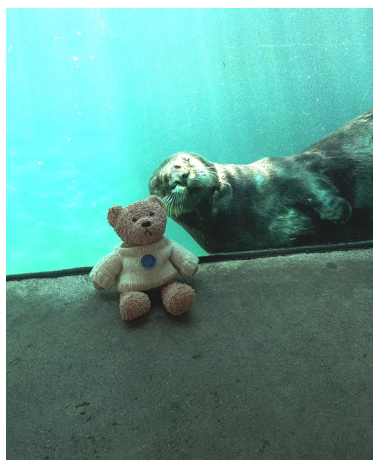


Caleb, Jasmine and Jordan. *Photo by Jasmine Champagne*

*(Continued on page 12)*

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

## A Barth Bear's Adventure *by Baby Barth Bear and Robert Hope, 33, Barth Affected Individual*



BBB at the aquarium.  
*Photo by Robert Hope*

A number of years ago my parents received a special stuffed Barth bear, quickly named 'Baby Barth Bear' (or BBB), and became the family mascot to take along on trips and help raise awareness about Barth syndrome. After a few trips with my parents, which BBB claimed were boring, I decided to give him a few experiences that he otherwise would not have had. In 2018 we toured Ireland, making sure to include a well-known brewery and a famous whiskey distillery, and, in June of 2019, he joined me on my Canadian west coast trip. The following are BBB's thoughts and views of the journey as dictated.

"While flying is not my favourite mode of transportation, I was very excited to be going somewhere again—it has been very boring just sitting on the corner of the desk. After landing in Vancouver, I insisted that we take a walk through a beautiful park along the water before going to an

aquarium. At first, I thought looking at sea creatures would be tedious, but when an otter came along for a chat, things became much more interesting.

The main reason for this trip, was to go on a train ride through the Rocky Mountains—I have never seen anything that high before. It was the middle of June, and there was snow at the top, glad we were not hiking them! Being the social creature I am, as soon as we got on the train, I quickly made friends with Jasper Bear--who is now also sporting a BSFCa pin. Jasper has made the trip many times, and told us what to watch for. Sadly, while the scenery was impressive, we were not able to see any of our bear cousins walking in the wild.

After two days on the train, I was happy to arrive at our stop. We spent a couple of days in a small town in the heart of the Rockies looking at lakes and mountains, and more snow! Robert even felt that a bus tour to the icefields was in order. Unfortunately, the weather was so bad, the road was closed, and the bus had to turn around so we returned to our hotel for a day of rest.



BBB and Jasper Bear.  
*Photo by Robert Hope*



BBB at Lake Louise. *Photo by Robert Hope*

Robert then decided that we should finish our vacation with a road trip to some badlands and to go see dinosaurs. Upon reaching the Royal Tyrrell Museum of Paleontology, Robert really lost it—I mean letting me be eaten by an Albertosaurus and then trampled by a frilled dinosaur was not going to do either one of us any good, other than apparently give him a good laugh. After my escape, it was time to head home—maybe sitting on the desk isn't such a bad thing."

## An Interview with Travis by Shelley Bowen, Barth Syndrome Foundation

### What advice would the twenty-four-year-old Travis share with the younger Travis?

*Stay active. Move, just move; even if it's just to walk for two minutes or lift two-pounds, keep moving.*

### Are you active?

*Yes, I enjoy staying active. Todd Cade's research was personally meaningful to me because it suggests how important it is for people with Barth syndrome to stay active. When you have Barth syndrome you don't burn fat for energy like healthy people, you use more glucose. When I was little my cardiologist told me never to become a couch potato because it would be harder for me to get reconditioned than it would be for a healthy person.*

### What else do you enjoy doing?

*I enjoy reading. I started playing guitar at a very early age probably around the age of six or eight. It was something that came naturally to me. My dad played the guitar and taught me and both of my siblings to play. I took private guitar lessons for about six years, then I started teaching lessons. Music lessons were fun, I guess because music came natural to me. It wasn't like school.*

### What do you remember about elementary school?

*Not being there. I missed a lot of school. I was sick a lot when I was young. I wasn't around the kids in school on a daily basis. That made it hard to fit in when I was there. They already had their friends. I guess I just didn't really feel like I could relate to other kids in school because they were not a part of my everyday life. Thankfully, I had many adult neighbors and friends in my life who were a constant in my life.*

### How do you feel activity is different now than it was when you were younger?

*Now I tend to know my limits. That took a while. When I was in physical education in school, I was being expected to do the same thing others were doing and I couldn't. I'd be pushed or push myself too far and it would make me physically ill. It's no fun to vomit in front of the entire school. One of the worst experiences I ever had was with a beep/bleep test in PE. That was brutal. I heard that it was banned because it was inhumane. If it wasn't, it should be.*

### Is there something that happens or a feeling that you get when you feel you are getting close to that threshold of no return with physical activity?

*I got sick many times before I found that line. I wanted to go and still want to go as far as I can. It's really important to me to remain active. But it definitely sucks to push beyond that line because if you do you are out for the whole day. It still happens, but not as often as it did when I was younger. It tends to happen more during the winter or right after winter when I am not as active. I feel better when I am active. I have to move every day. Small things add up and help me to feel better. Inactivity adds up too, only not in a good way. I'm not suggesting that I am ready for a 5k run but to me it's not about pushing myself to reach the standards of others, my limits are personal.*

### What was school like for you after elementary school?

*I left school in the eleventh grade and went back about a year later to the adult educational center. I had*



Travis. Photo by Amanda Clark

(Continued on page 8)

### Virtual 5K Run *by Chris Hope*

To welcome spring, we are going to have a virtual 5 Km run fundraiser. It is simple and easy, and can take place at any location you choose, anywhere in the world. Registration is \$50.00, and is open from Feb 29<sup>th</sup> to April 18<sup>th</sup>. Prior to May 1<sup>st</sup>, you will receive a custom race medal, then anytime during the month of May, you get to run your race—you can run, jog, or walk on the road, on the trail, the treadmill, on a track, or even at another race. You can even bike, if you prefer. You can run at your own pace—all at once, or over as many days as you like. When completed, take a picture with your medal and share with us on our run Facebook page ([www.facebook.com/groups/177156293658074/](http://www.facebook.com/groups/177156293658074/)).

All proceeds go directly to BSFCa, and will help us continue to work towards finding treatments and a cure for Barth syndrome.

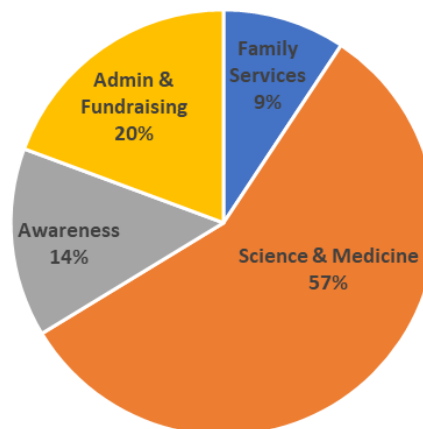
To register, please go to our event fundraiser page: [www.canadahelps.org/en/charities/barth-syndrome-foundation-of-canada/events/virtual\\_5\\_k\\_run](http://www.canadahelps.org/en/charities/barth-syndrome-foundation-of-canada/events/virtual_5_k_run).

### 2019 Finance Report *by Chris Hope*

Our purpose and mission as a charity is to enhance the lives of those affected by Barth syndrome. We use a three pronged attack to achieve this—we provide information to physicians and health care workers so they can make the diagnosis quickly, and then treat their patients accordingly, we educate and support the families and individuals so they know how to best care for themselves, and we fund Barth specific research to find better treatments and eventually a cure.

We remain a completely volunteer foundation and rely solely on donations and grants from generous and caring individuals and corporations. We were able to offer last year's (2019) outreach through the benefit of a grant received from the Max Bell Foundation.

Where the Dollars Go



# Barth Syndrome Foundation of Canada Virtual 5k Run/Walk



**MAY 2020 RUN OR WALK ANY DAY, ANYTIME, ANYWHERE!**

## Registration February 29 - April 18

[https://www.canadahelps.org/en/charities/barth-syndrome-foundation-of-canada/events/virtual\\_5\\_k\\_run](https://www.canadahelps.org/en/charities/barth-syndrome-foundation-of-canada/events/virtual_5_k_run)

\$50 per person with proceeds going to Barth Syndrome Foundation of Canada. Registration includes custom race medal. Tax receipts issued according to CRA regulations.

**Join us on Facebook for event updates, to learn more about this rare disease and to meet some of the amazing guys affected by Barth Syndrome.**

<https://www.facebook.com/groups/177156293658074/>



Barth Syndrome Foundation of Canada

**Contact Jasmine at [champagne.jasmine@gmail.com](mailto:champagne.jasmine@gmail.com) for more info.**

Barth Syndrome Foundation of Canada  
162 Guelph St., Suite 115  
Georgetown, ON L7G 5X7  
[www.barthsyndrome.ca](http://www.barthsyndrome.ca)  
905.873.2391  
Reg. Charity # 86102 2002 RR0001

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

## A Fun and Heart-Warming Weekend *by Chris Hope*



Barth Guys Celebrating Star Wars Day.  
*Photo by Susan Hone*

Last year, it was decided to expand our annual outreach to a weekend event instead of just one afternoon allowing us to be able to plan a varied agenda—one that included social time as well as information.

We were pleased to provide several speakers who gave presentations and answered questions.

Dr. Matt Toth, BSF Science Director, brought everyone up to date on the most promising research options, and explained the next steps and hurdles. Dr. Robin Duncan, a researcher from the University of Waterloo, talked about an exciting new discovery from her lab, which she is continuing to investigate,

and Dr. Iyar Mazar, who has worked extensively internationally with both young and older individuals with Barth syndrome, spoke about her work on the experiences of those affected with the disorder—the management of impact the disease has, as well as means for goal setting in tenuous health states.

Following this, families and affected individuals were able to have a discussion on some of their challenges and goals for the future.

We heard about Sheldon heading off to university this fall, Ryan and his fiancé Jessica spoke about their wedding plans, and Adam related his first experiences about moving out on his own.

The young men made it clear that they are ready to live their lives their own way, and that ‘they are fine, thank you very much’.



Group photo of the 2019 Outreach. *Photo by Susan Hone*

Everyone was able to join in a family-style dinner, which was then followed by an enjoyable evening of socializing and mingling.

Sunday morning saw several spirited games of bowling followed by a pizza lunch, after which it was time to head home.

A fun weekend for all, made more so with the addition of families and affected individuals from the US.

## 2020 International Conference by Jasmine Champagne

Barth syndrome is rare, life threatening, and many times an invisible disease filled with unknowns and a life of uncertainties. The Barth Syndrome Foundation International Scientific, Medical, and Family Conference changes that, even if only for a little while. It is a place to learn about the latest research and potential therapies. It is a place to make connections, new friendships, and a new family. It is a place to be surrounded by others that know exactly what living a life with Barth syndrome looks like, a place to find love and hope.

The theme for this conference is “For a new GENERation”. Celebrating the past twenty years, and looking to the future with hope. As in the past, this year there will be research studies, family and youth educational sessions, and updates in research, science and medicine for researchers, clinicians and scientific professionals. There will be an emphasis on connection and community building, as well as a forum for healthcare providers on symptom and disease management.

Various committees have been hard at work for months ensuring that this will be the biggest and best conference ever.

At the conference, a unique relationship is created between researchers, affected individuals and their families, and clinicians. It is this open and exceptional collaboration that will facilitate the discovery of treatments for Barth syndrome.

To learn more and to register, please visit: [www.barthsyndrome.org/conference](http://www.barthsyndrome.org/conference).



Individuals affected by Barth syndrome at the 2018 Conference. Photo by Jasmine Champagne

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

*An Interview with Travis by Shelley Bowen, Barth Syndrome Foundation*

(Continued from page 3)

*gone through my growth phase. People had their own unique challenges. They were there because they wanted to be. Your education was on you. They worked with me, but ultimately it was on me to do the work. Things change. I learned at a different pace at some point. I didn't feel as sick everyday as I did when I was younger. Maybe that's why I was able to learn. After graduating I went to work but it wasn't a good fit for me. So, I went back to school.*

**What did you study?**

*When I went back to school. I chose an area of study that fit me. I had some work experience to know what I liked, what I didn't like, what I could do and what was too much for me to do. I also worked in sales and learned that wasn't for me.*

*I completed studies for developmental services to help people with developmental disabilities. But that really wasn't a good fit either.*

*When making a decision about my future, I considered my health a lot. I'm doing well now. I don't take that for granted. My health could worsen at any point. So, I have to consider the level of debt I might have with going back to school, how much time out of my life would be focused on school and would it be worth it? I don't want to go to school for eight years and be in such debt that I have to work sixteen to pay it off. What if I can't? Ultimately, it comes back down to having the training to do a job that will be a good fit for me.*

*I am going back in the fall for speech and language pathology. I can't sit at a desk all the time. It wouldn't be good for my health. I can't be in a job that is too physically demanding because that wouldn't be good for me either. I have to find a career that will support me and maybe other people, but it has to be a job that I am good at and something that I can keep doing for a long time.*

**Do you want to have a family in the future?**

*Yeah, I do. I think about it. I'm at the age where I am beginning to think about that more. Who knows what will happen?*

**Does Barth syndrome factor into that decision too?**

*I guess so, I mean the whole "Do I want to have a child who might be a carrier" is something I have considered. It's not like Barth syndrome is keeping me from starting a family but it is something that has to be considered.*

**When did the reality / seriousness of Barth syndrome become real to you?**

*Well, there are those little reminders that it's there like when I push myself too far. It's like "Oh there you are." There was no specific event that I can cite. Missing a lot of school when I was a kid and going back and being different was a reality check. Getting sick in front of my classmates was a reality check. Wondering if I am going to be around to pay off my school debt gives me pause. When a friend who has Barth syndrome dies, that's a reality check. Barth syndrome is like that sneaky little devil on your shoulder that taps to remind you it's still there. It's not just one thing, it's the constant reminders that you need to learn how to handle.*

**Travis's Mom's Perspective**

*It has been a journey. It was really hard when he was little in school. It was a fight every day to get him to go to school. He dreaded it and so did I. I am very proud of him. He loves life. He is curious. He made it through college and now he's going back to university. I will always remember how difficult it was when he was in school. It was hard because he looked so well. No one took me seriously when I told them about the seriousness of his condition.*

(Continued on page 10)



### Research Update by Chris Hope

Since 2006, BSFCa has been contributing as much as possible to research, as this is the only way that we will ever be able to find effective treatments and ultimately a cure for Barth syndrome. After many years, and even more ideas, researchers have come up with three promising potential new therapies to dig deeper.

- Gene Therapy
- Enzyme Replacement Therapy
- Modifier Gene ALCAT1

In gene therapy, the idea is to add a functional TAZ gene (the cause of Barth syndrome) while with enzyme replacement therapy a protein would be given to an affected individual. On the other hand, ALCAT1 therapy relies on a different gene to modify the function of the TAZ gene.

While much work still needs to be completed before any of these ideas can be brought towards a clinical trial, the news is encouraging, and we are all waiting to hear more.

### A Rare Opportunity by Susan Hone

On June 18th, 2019 Christiane Hope and I gave a 40-minute presentation on Barth syndrome entitled *From Tears and Fears to Advocacy and Hope* at the Canadian Human and Statistical Genetics Meeting/ GE3LS HSPR\* Conference 2019 in Montebello, Quebec. We, Barth Syndrome Foundation of Canada (BSFCa), were invited to speak by Chris McMaster, PhD, FCAHS, Scientific Director, Canadian Institute of Health Research (CIHR) Institute of Genetics and Professor of Pharmacology, Dalhousie University. Dr McMaster has attended two Barth Syndrome Scientific, Medical and Family Conferences and was a Barth Syndrome Foundation, Inc. Research Grant recipient twice. BSFCa funded \$18,000 USD in the 2007 grant cycle for *Synthetic genetics toward understanding Barth syndrome cell biology*, and \$40,000 USD in the 2010 grant cycle for *A screen for drugs leads for the treatment of Barth syndrome*.

A few of the conference features were advocacy groups and individuals telling their stories of living with a rare disease, the Clinical Implementation of Genomics/Genetics, Health Policy Research and Rare Disease, a Health Canada workshop in which we participated in discussions on the key considerations Health Canada should consider for the development of a regulatory sandbox and how they can ensure that Canadian interests are balanced. We made connections with other rare disease organizations and spoke with researchers about the importance of their work. While several physicians told us that they had heard of Barth syndrome, and even sent away for testing, no one had to date met anyone with the disorder.

The three days went by quickly and we came home with a list of websites to check into further and new contacts to help us raise awareness about Barth syndrome.



Chris, Susan, & Dr. McMaster.  
Photo by Susan Hone

\*Genomics and its Ethical, Environmental, Economic, Legal, and Social Aspects; Health Services & Policy Research

## Friends, Cars and a Spectacular Birthday Party *by Lynn Elwood*



Adam and Bob.  
*Photo by Lynn Elwood*

When Bob McJannett holds a birthday party, it is a day like no other! The McJannett family have been terrific friends of Barth and have held fundraising events several times over the years. Their backyard party fundraisers have become a highlight of the year, bringing friends from near and far to share in an afternoon of friendship, music, refreshments and some amazing cars.

Despite having a challenging year, Bob opened his home to his friends and held an incredible birthday celebration. Once again, Bob used the opportunity to raise funds for Barth syndrome, bringing in over \$5000 that will help tremendously to fund our programs such as research, awareness and family support.

This year was the first time we had an affected Barth individual join the party as Adam Elwood came to the event.

The day was perfect, and we were greeted by some fantastic cars, including an antique car out front of the party and several Mustang convertibles. Adam and the other car enthusiasts had a terrific time sharing stories and admiring the “auto show”.

It was a pleasure to spend time with Bob and his sons, and their amazing group of friends. There was great music with Johnny Max Band, a bit of dancing, and lots of delicious food. And most of all, there was time with great people. It was a very special afternoon full of caring, camaraderie and laughter. We cannot thank Bob and family enough for holding this event, and for supporting Barth Syndrome Foundation of Canada through their generosity. Happy Birthday Bob!



*Photo by Lynn Elwood*

## An Interview with Travis *by Shelley Bowen, Barth Syndrome Foundation*

*(Continued from page 8)*

### **How would you describe your Barth syndrome journey to others?**

*I'm lucky, I'm really lucky. That might change, I hope not. Right now, things are good for me and I am very thankful for that. I'm also very thankful that I have been able to stay active. I hope that doesn't change.*

### **In Appreciation** by *Chris Hope*

The Barth Syndrome Foundation of Canada is entirely run by volunteers, we do not receive any government funding, and rely completely on individuals' and corporations' donations and fundraisers. Because we are a rare disorder, we are not a household name, every time someone shares or mentions Barth syndrome, it increases our circle of friends. We appreciate every researcher who shows an interest, every physician who treats and cares for individuals with Barth syndrome and shares their experience, every family member and friend who comes to an event, everyone who holds and participates in a fundraiser. We are grateful to the individuals affected by Barth syndrome; they participate in research studies and trials and support and encourage each other. We thank their friends and siblings who also participate in the studies as controls. There would be no progress without everyone pitching in. Every contribution and gift makes a positive impact on those affected by Barth syndrome, and we are truly grateful.

### **Glass Turkey Fundraiser** by *Lynn Elwood*

What is a Glass Turkey? It is a basket full of adult beverages of all types. It is also a great prize in a fundraiser that was held in the fall of last year. Cathy Ritter's brainchild, this fundraiser had us selling raffle tickets for a chance to win one of two Glass Turkeys, just in time for the holidays. Thanks to Cathy and Lynn for holding this personal fundraiser that brought in over \$1600 – and to Chris, Lois, Robert and the others who bought and sold tickets, and donated the prize contents. Congratulations to Keith Leighton and Sienna Morris who won the baskets.



Adam, Les, and Rick at Bob's birthday party.

*Photo by Lynn Elwood*



Glass Turkey Fundraiser with Ryan and Cathy Ritter.

*Photo by Cathy Ritter*

## President's Report *by Susan Hone*

*(Continued from page 1)*

aboard Jasmine!

Another volunteer who is tirelessly working for us behind the scenes is Paula Sisson. She continues to update our website and produce our newsletter. Thanks for all the hard work Paula!

In May, we said farewell to the first Science Director employed by Barth Syndrome Foundation, Inc., Dr. Matt Toth. Matt was instrumental in finding researchers who are as devoted to finding a

treatment/cure as Matt was. We welcome Dr. Erik Lontok as the new Director of Research and wish him success in his new role. Chris Hope and Susan Hone were fortunate to meet Erik at a planning session for the upcoming BSF Scientific, Medical and Family Conference being held in July 2020.

BSFCa has supported the BSF conference for a number of years. This year is a major milestone for the conference as it is the 10th conference and 20 years since a group of parents and individuals with Barth first gathered together. A number of Canadians are volunteering to help make this conference the biggest and best to date. As well, BSFCa will be sponsoring portions of both the scientific and family side of the conference. Hope to see a number of you there.

As always, we are very thankful to our donors and volunteers. Without you, the above events and research could not happen. Thank you for choosing Barth Syndrome Foundation of Canada as one of your charities of choice. You are changing the lives of individuals and families affected by Barth syndrome.



Dr. Matt Toth with Barth Individuals *Photo by Susan Hone*

**For online donations, please visit [www.canadahelps.org/en/dn/3811](http://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 *tafazzin* 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](mailto:info@barthsyndrome.ca)

Barth Syndrome Foundation of Canada, 162 Guelph Street, Suite 115, Georgetown, ON L7G 5X7  
(905) 873-2391, 1-888-732-9458, [info@barthsyndrome.ca](mailto:info@barthsyndrome.ca)