



Barth Syndrome  
Foundation of Canada

# Canadian Newsletter

www.barthysyndrome.ca

November 2016



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Barth Syndrome  
Foundation of Canada

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



## President's Report by Susan Hone

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 8<sup>th</sup> 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 to the Barth syndrome dream. Thank you to all who are now a part of my world-wide Barth family.

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.

With our fund-raising efforts, BSFCa was able to contribute to the conference by sponsoring the photo booth, poster session, a breakfast and three Canadian physicians/ researcher's flights and accommodations. One of these recipients, Dr. Robin Duncan, Assistant Professor, University of Waterloo, Waterloo, Ontario was awarded a grant partially funded by BSFCa entitled [A new enzyme and pathway in cardiolipin synthesis](#).



Florence Mannes, Chair, Association Barth France; Susan Hone, President, Barth Syndrome Foundation of Canada; Mark Sernel, Chair, Barth Syndrome Foundation; Michaela Damin, Chair, Barth Syndrome Trust; Paola Cazzaniga, President, Associazione Barth Italia. Photo by John Wilkins

We also sponsored \$1,000 to The Biophysical

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## 2016 Conference: Therapeutic Ideas Take Center Stage

*by Matthew J. Toth, BSF Science Director*

Focusing just on the Science and Medicine track of the conference, it was remarkable to hear about an overwhelming number of potential and ongoing therapeutic ideas and therapies being considered and being implemented. Over eight therapies were discussed at the SciMed sessions, and one biotech company presented their clinical trial plans to the Scientific and Medical Advisory Board for input. Therapeutic ideas were discussed that ranged from traditional pharmacological ideas, to biologics, to gene therapy. Several of these therapeutic ideas are so cutting-edge, that there is little or no precedent for them.

I had the privilege of sharing what I knew about these therapies with many of the Barth guys on Monday evening over pizza (which tasted pretty good) before the SciMed sessions. Over the next two days I met primarily with parents to give the same talk. There has been nothing more gratifying in my professional career than to give those presentations! In July 2006, after just starting with BSF two weeks before, I attended my first BSF Conference where I spoke to the parents about having a therapy for Barth syndrome. It took 10 years, but I had the good fortune to talk about eight therapies in 2016. Now my message to the Barth guys and their families is about their need to volunteer for the clinical studies and clinical trials that will be coming.

“Team Barth” was the motto of the 2016 Conference because it will take a team effort to get BSF to the next and final level. We need Barth guys to volunteer for clinical studies so that the promises of 2016 do not evaporate. At the Monday evening pizza meeting with the Barth guys there was a point where one of the older guys came in late, most likely due to the fatigue and pain that so crushes him and others. He spoke about how important it was for him to be able to help his “brothers”, how great it is that he could volunteer for the “next big thing” that has the potential to improve their lives, and about how all of them need to give their best effort so that the next generation of Barth guys, and maybe even themselves, will live better.

Through the BSF Research Grant Program and these biennial Conferences, BSF is now “reaping” what they began “sowing” back in 2000. BSF has a cadre of dedicated researchers and clinicians pushing the limits of science and medicine to deliver what we all want—effective treatment for Barth syndrome. The SciMed presentations are remarkable for their openness and critical discussion. It is not often that researchers feel comfortable about putting up their latest results for scrutiny, warts and all, for discussion with their peers. But at the BSF Conference this openness happens consistently. Scientists and clinicians are ordinarily cautious and conservative in their remarks in public, and to hear and see them interact in such an open way with each other is refreshing and actually exciting. Through its culture, the nature of the disease, and maybe the guys themselves, BSF has been rewarded with a great group of health professionals and researchers.



Dr. Matthew Toth.

*Photo compliments of BSF*

It is hard living with a rare disease that has no specific treatment. It is also hard to realize that the final steps in doing something about a rare disease can only be done by those who suffer from it. At this meeting one could see a glimpse of only some of the crushing weight that Barth guys have to live through, and now we have to ask them to give another full measure of effort. We ask them because no one else can do it. Too bad we cannot spread around the burden to others. Like the disease itself, volunteering for clinical studies is something only the Barth guys can do or have to endure.

At the Family sessions of the conference, talks by Marc Sernal and Kate McCurdy really resonated with the group as everyone realized that the future they dreamed about was finally coming to fruition.

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## Personal Perspectives of the Barth Syndrome Foundation Conference

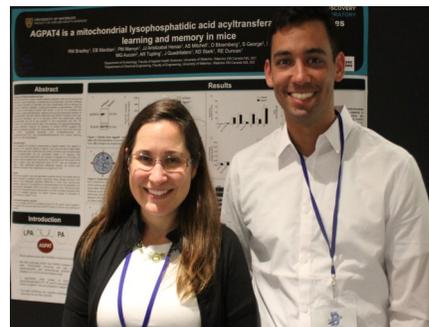
by Dr. Robin Duncan

In my scientific career, I have never experienced anything like the Barth Syndrome Foundation 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’s 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 Barth Syndrome Foundation and Barth Syndrome Foundation of Canada for such kind support, and for an experience that left me both optimistic and energized.

*Dr. Robin Duncan is an Assistant Professor of Physiology and Nutrition at the University of Waterloo, Waterloo, Ontario. This was her first time attending the Barth Syndrome International Scientific, Medical and Family Conference. Dr. Duncan was invited to present her work at the Science and Medicine Sessions.*



Dr. Robin Duncan and research student Ryan Bradley at the BSF Conference. Dr. Duncan was sponsored by BSFCA to speak at the conference. *Photo by Susan Hone*

## 2016 Conference: Therapeutic Ideas Take Center Stage by Matthew J. Toth

*(Continued from page 2)*

The conference clinic was remarkable for the number of studies and the quality of the research that was being performed. The pill-swallowing workshop was particularly special for the success the Barth guys had in performing this extremely important skill that is difficult for them. Other talks that dealt with the problems and some solutions for living with Barth syndrome were well received.

From the beginning to its end the 2016 Conference was fabulous. Both the family group and the scientists, researchers, and physicians group enjoyed themselves and learned a lot. It was a turning point for BSF, where we stopped talking about therapies, to one where we have to do something about therapies. It is a brave new world for sure.

### **From the Heart** by *Cathy Ritter*

“Life isn’t about waiting for the storm to pass – it is about learning to dance in the rain.”

~Vivien Greene~

It has been many years since our family learned about Barth Syndrome. With the diagnosis came mixed feelings. It helped explain many unanswered questions regarding three precious baby boys who were no longer with us. It also brought fear – for the child I was expecting, for my other two children and also for my cousin’s son who had been diagnosed with BTSH.

On May 16, 1991 our youngest son Ryan came into the world. He was big, pink and had a lusty cry. He was checked out from head to toe by the paediatrician at the hospital – we were told everything looked good. At the time there was no genetic test for Barth Syndrome, so when only a few weeks old, we made the trip to Sick Kids for Ryan to have his heart checked out. At that visit, we were told that Ryan did indeed have Barth syndrome. The storm we had been living for the past few years swelled into a torrential downpour.

During the early years we learned to live with BTSH. Yes, Ryan had many tests, supportive therapies and was seen by many “ologists”. He also played baseball, started school, went to cubs and was bossed around by his older siblings. Although BTSH was always present in the back of our minds, we were determined that he was going to live the “regular life” of a little boy. Throughout those years other families around the world were also learning about Barth syndrome. In 2000, several of those families gathered together and the Barth Syndrome Foundation was born. In 2002, Ryan and I along with my cousin and her son, travelled to Baltimore to attend the first Barth Syndrome Conference. Words can not really express the experience – for the guys to meet others just like them, for the parents to talk to someone who really does get it, to have specialists there who know BTSH ... it was a totally amazing experience and a whole new world opened up. Attending the bi-annual conferences has been a priority for Ryan and I. It affords an opportunity to continue friendships, to feel connected to the Barth family community and to participate in the research dedicated to finding treatments and therapies for BTSH. One of the highlights of the conference is the Friday night social. It is a truly special evening with everyone together: friends, families, doctors and scientists. It is an evening of hope and promise... and dancing - no matter what the weather.

A few more years have passed and Ryan is now 25. He graduated college with a Diploma in Culinary Management and has been working as a chef for several years. He is currently living in London, Ontario with his girlfriend Jessica... and living the life of a regular young man.



Cathy and Ryan Ritter - BSF Conference 2014. Photo by Amanda Clark

## Barth Syndrome Foundation of Canada (BSFCa) Planning Session 2017 by Susan Hone



BSF Executive Back: Susan Hone, Carol Wilks Front: Lynn Elwood, Chris Hope, Lois Galbraith. Missing: Cathy Ritter. Photo by Susan Hone

The 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 Kashabog, Ontario. Attending were Lynn Elwood, Chris Hope, Susan Hone, Lois Galbraith, Carol Wilks and our chefs/dishwashers, Les Morris (also Board advisor) and Adam Elwood. 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 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. Once 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.*

### Giving Tuesday – November 29, 2016

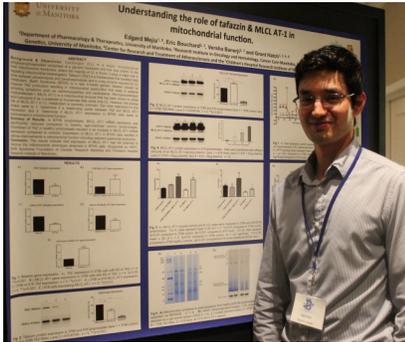
Giving Tuesday has become a global day of giving back. This is a special day set aside to give everyone the chance to join people around the world to support a cause close to your heart.

We would be very grateful if you remember us on this giving Tuesday to help us give those with Barth syndrome the chance of a better life. To make an on-line donation, please visit our website at [www.barthsyndrome.ca/donate](http://www.barthsyndrome.ca/donate).

We are excited to announce that a generous donor has stepped up to **match the first \$2,000 raised**. This gives you the chance to make **TWICE** the impact.

Mark your calendars and spread the word!

## A Return Experience by *Edgard Mejia*



Edgard Mejia with his poster submission at the BSF Conference. *Photo by Susan Hone*

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 is a Post-Doctoral Fellow, Department of Immunology at the University of Manitoba. He has attended two BSF conferences and submitted posters each time..*

## Conference Reflections by *Ryan Bradley*

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 8<sup>th</sup> annual Barth Syndrome Foundation conference this past July, and it was truly an unforgettable experience. Due largely in 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



BSFCa Directors Susan Hone and Lynn Elwood with researcher Ryan Bradley at Poster Session funded by BSFCa at BSF Conference. *Photo by Susan Hone*

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### **My Perspective of the Conference** *by Adam Elwood*

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 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 (a BSF volunteer) 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 new/young guys with Barth 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!!



Adam Elwood. *Photo by Lois Galbraith*

### **Volunteer Corner** *by Lois Galbraith*

We love our volunteers – they are the ‘heart beat’ of the foundation. Our volunteers are skilled, dedicated and eager!

The only thing better than showing our appreciation to our volunteers is actually ADDING to our volunteers.

We introduce two new volunteers, a mother and daughter team, Cheryl and Jeannine Leighton. They joined our team to help with a sewing project and produced some of the blue medical pouches for families at the BSF Conference this July.

Jeannine has also been completing web searches for us. She is helping us to narrow down lists of ‘grant-giving’ corporations. Her work for the BSFCa pays off for us at her workplace as well. Each year that she volunteers forty plus hours for us, her employer (RBC) gives us a grant of \$500.00. Another win-win situation for us!!

Welcome Cheryl and Jeannine!!



Jeannine and Cheryl Leighton. *Photo by Lois Galbraith*

## **President's Report** *By Susan Hone*

*(Continued from page 1)*

Society of Canada's conference in June. This was the first medical conference in Canada, which we are aware of, where Barth syndrome was on the agenda. Dr. Richard Eband chaired a session related to Barth Syndrome/Cardiolipin and Mitochondria. Among the speakers were doctors Michael Schlame, Valerian Kagan, Grant Hatch, Miriam Greenberg and Stephen Claypool.

On the home front, we had a family activity in September along with our annual planning meeting. We have a volunteer scouring the internet looking for grants we can apply for to raise funds. We will again be participating in Giving Tuesday as we had great success raising funds during last year's event. 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 can not 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.

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## **Conference Reflections** *by Ryan Bradley*

*(Continued from page 4)*

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 will continue to wear it until the end of my doctoral studies, at the very 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 is a PhD Student – Physiology & Nutrition at the University of Waterloo, Waterloo, Ontario in the Faculty of Applied Health Sciences and presented two abstracts at the Barth Syndrome 8<sup>th</sup> International Scientific, Medical and Family Conference Poster Session.*

### **A Day with my Barth Family by Sheldon**

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 the 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 like 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.



Sheldon, Travis, Adam, Jared and Robert. *Photo by Susan Hone*

### **Memories of the 2016 Barth Conference by Susan Hone**

*(Continued on page 10)*



Our group of boys, young men and gentlemen who are affected with Barth syndrome. *Photo by Susan Hone*

## Memories of the 2016 Barth Conference by Susan Hone

(Continued from page 9)



Luminaries at night, set up by the Hone family and many other volunteers. They represent those living and deceased affected with Barth syndrome. *Photo by Susan Hone*



Chris Hone, Lois Galbraith and Josh Hone at the Saturday evening social. *Photo by Susan Hone*



Intensely listening to one of the presenters. *Photo by Susan Hone*



BSFCa Sponsorship sign. *Photo by Susan Hone*



Lynn Elwood, Barth man aka Chris Hone and Florence Mannes, Chair of Association Barth France. *Photo by Susan Hone*



Being piped in to light the luminaries. *Photo by Susan Hone*

## Friends of Barth

1586732 Ontario Limited	Haggett, John & Judy	Perkins, Phyllis
Alblas, Henk	Hammond, Jean	Pickup, Amanda
Allen, James	Hatch, Grant	Pitkethly, Maureen
Allison, Vicki & Gord	Herner, Brian & Barbara	Poitras, Mike & Sandi
Barrett, Lawrence & Marilyn	Higley, Hazel	Prodan, Magda
Basilio, Anita	Hintze, Audrey	Quackenbush, Earl & Carolyn
Best, Cathy & Rance	Hone, Barbara	Raymond, David & Lila
Best, Riley & Jennifer Hone Best	Hone, Chris & Susan	RBC Foundation
Bohns, Hannelore & Alfred	Hope, Emily	Ritter, Cathy & Chris
Bradley, Ryan	Hope, Harry & Helen	Robertson, Heather
Bridger, Wayne & Dianne	Hope, Michael & Christine	Ross, Barbara
Burmania, Owen & Sharon ~	Hope, Robert	Segal, Donna
Sunsational Landscapes Inc.	Hope, Terry & Ruth	Segal, Joseph
Butler, Gayle	Howard, Sharon	Segal, Susan
Chambers, Ronald	Hucaluk, Adrienne	Sheridan, John ~ ScotiaMcLeod
Cherniak, Andy	Humphries, Jack & Jan	Sills, Brenda ~ Carlson Wagonlit
Cherniak, Leah	James, Jennifer & Ryan Ritter	Sisson, Paula
Cherniak, Miriam	Jeffery, Elaine	Storey, Dick & Janis
Clelland, Bill	Kugelmann, Jan	Sturup, Penny
Conlin, Patricia	LaVigne, Carol	Timleck, Barry
Cooper, Jocelyn & Gerry	Leighton, Cheryl	Toth, Matt
Coulson, Steve	Leighton, Keith & Jeannine	Trim, Norma & William
Cowper, Ron	Lindsay, Sue	Trueman, Carole
Cummings, Marie & Jim	Loos, Ann	van Dam, John & Marian
Daley, Sheldon.	MacDonald, Fred	Vanderwater, Barbara
Davidson, Alex & Bonnie	Maier, Charles & Carol	Vella, Betty
DeForest, Wilmer & Ina	McGill, Sandi	Vine, Edward & Rosa
De Graauw-Bailie, Arlene	McGill, Steve	Warren, Jerry & Janet
Deligiannis, Dino	McGlaughlin, Larry & Jackie	Weller, Sharon
Dickson, Bel	McJannett, Susan & Bob	Wilks, Carol & Bruce
Doherty, Jerry	McKee, Jim	Wilks, Dennis
Dove, Judy & Randy	Megelink, Jasper & Mary	Wiwcharyk, Lubow
Duncan, Robin	Mejia, Edgard	Woodcock, Roy
Ebata, Lyle	Millar, Ronald & Margaret	Worsley, Dorothy
Elwood, Adam	Milne, Peter	Wu, Jin
Elwood, Bryan & Susan	Miloff, Maury	Young, Ron & Lenora
Elwood, Lynn & Rick	Miloff, Michael	Zavitz, Peter
Ernst, Angelika	Morris, Les & Lois Galbraith	
Ernst, Gabriele & Kevin	Murray, Allan & Jane	
Forbes, Carolyn	Nagel, Siegfried & Erna	
Fraser, Wendy	Nagel, Thomas	
Gallacher, Betty	Noddle, Bev & Norm	
Gilmour, John & Christine	Olson, Sharon	
Ginou, Jonathan	Paczkowski, John & Vicki	
Gordon, Karen	Perkins	
Gordon, Travis	Patrick, Barbara	
Haggett, David & Sharla	Pearce, William & Bev	



Volunteers filling  
luminary bags.  
*Photo by Susan Hone*

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### Taking Part in Dr. Cade's Research by Adam Elwood



At the end of October, I took part in Todd Cade's research study and got a new appreciation for how much time, money and effort is involved in the research studies being done to help the Barth guys. Going into the study I was a bit nervous. I thought it would involve long days, very tiring days, and it would be unpleasant.



I was wrong. There were lots of tests, but the days were not very long. The tests were mostly pretty easy - boring at times but easy. The researchers and the nurses really worked hard to make me comfortable and to keep things streamlined. When I was cold, they put warm blankets on me. When I was bored, they brought in a TV and movies to keep me occupied. They explained everything they were doing and were happy to answer all my questions.

Thank you to Todd Cade, Dominic, Adam and, most of all, to Kay Bohnert who was amazing and there for every part of the week. It was overall a nice week and I would encourage others to take part in the research any chance they get.

Adam Elwood in St Louis for Research Study. Photo by Lynn Elwood



## 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 Lois Galbraith at info@barthsyndrome.ca