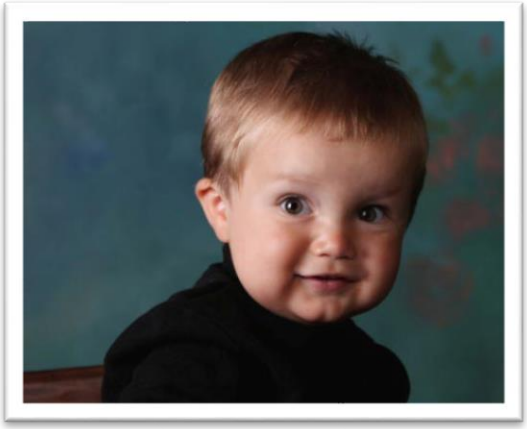
Barth Syndrome Foundation®

Quote of the Day:

"We live in a country where fewer than ten boys have been diagnosed. Yet, we never feel alone in facing our challenges with having a child with Barth syndrome because the Barth Syndrome Foundation is there to provide educational information, medical support and personal support when we need it. Even though we know a cure is not right around the corner we have confidence in knowing research is being conducted and we trust the Barth Syndrome Foundation to propel research, which will improve the future for our children." *~ Florence, Mother of Diagnosed Son, France*

Featured Story:
A Personal Message from Raphaël's Family



Raphaël

Raphaël is a cute 22-month old boy. Although he looks like a one-year old, he acts and understands as a boy of his age, which can be confusing sometimes.

He loves playing with his brothers, and is a very loving boy. His heart function is in the lower normal range, and looks, as many Barth syndrome (BTHS) boys, perfectly healthy. His big cheeks and his wide eyes make people stop in the street to tell us how cute our boy is….they cannot imagine how precious he is to us….

Raphaël was born on December 9, 2008. He is the third of our boys, born smaller than his brothers, Romain and Victor, but in good shape.

When Raphaël was one month old, we had to go to the ER for an unexplained fever. The doctors found out that his white blood cell count was low and that something was wrong with his heart. Two months later, Raphaël was diagnosed with dilated cardiomyopathy.

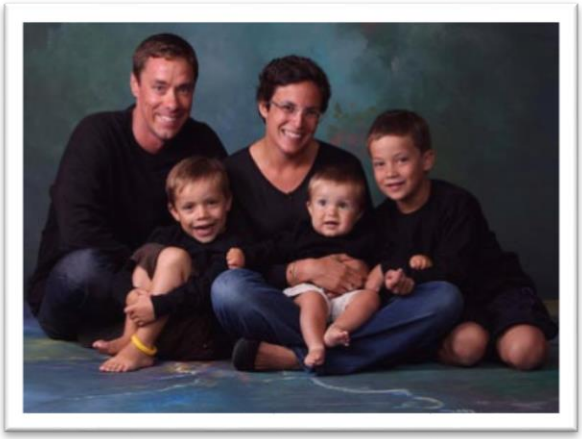
Cardiomyopathy, together with neutropenia, rang a bell with Raphaël's cardiologist, who requested that Raphaël be tested for BTHS. We had to wait five months before the results of the diagnostic test were available. This was a very long period of time, not knowing what to expect and not being able to look in any direction to get help. We felt we were stuck in the middle of nowhere.

Results from the test finally came on August 19, 2009. We will never forget this date. We couldn't consider that this diagnosis was good news. We already knew this disorder still claims the lives of many boys, but, strangely, we felt a bit relieved, since we were not alone to fight.

During all the time we were waiting for a diagnosis and also the very first days after Raphaël was diagnosed, we spent a lot of time on the Internet trying to find as much information as we could. Most of the information we gained was from the Barth Syndrome Foundation (BSF) website, which was so helpful. We began to learn more about the medical aspects of the disease, and we also learned about the everyday life troubles and how to cope with low energy, gross motor delay, eating, etc.

Only a few days after we got the diagnosis, we sent an e-mail to ask how to be part of the Foundation. Shelley Bowen replied to our e-mail almost immediately and included us on BSF's Family listserv. We were also put in touch with Michaela Damin and Annick Manton of the Barth Syndrome Trust (BST). Annick is responsible for Barth Syndrome Trust's family support, including families in Europe and has continued regular contact with us.

Welcoming e-mails were so warm, information was so useful, and we were amazed to see that we were not alone and that many families around the world were facing the same problems. This was such a surprise for us. It felt as if we had a new family who was able to understand what we were experiencing with Raphaël.



(L-R) Phillippe, Victor, Florence, Raphaël & Romain

Last January, we decided to attend BSF's 5th International Conference, which was held in July in Florida. It was, for our kids, their first "big" trip; and it was, for us, the first time we met other BTHS families who already felt to us as friends after reading their e-mails on the listserv. Even if it could be hard sometimes to meet parents that have lost their sons and to face what BTHS could mean, this conference was so full of hope. We met boys, parents, doctors, family members, all of them dedicated to one single goal: helping these boys, to which the sentence of Gandhi applies so well: **"Strength does not come from physical capacity. It comes from an indomitable will."**

We were amazed at the commitment of those who organized the conference, and are willing to help. Back in Paris, we decided to create an Association, called "Barth France," in order to raise money for BSF and also to raise awareness here in France. We are convinced that there are more than a couple of boys affected with BTHS in France…there are just doctors that do not know about the disease.

Philippe, his brother, Raphaël's godfather and three of their friends are now training for the Ironman France–Nice on June 26, 2011. Inspired by Gary Rodbell's conference presentation about his fundraising and awareness success associated with Ironman races in the US, we have been in close touch ever since. We hope to be able to raise money during this race and will run under the colors of BSF.

When we realized what some mothers (the very first ones to create the Foundation) had to face, and when we measure the progress made in 10 years, we are so thankful and our hearts are full of hope. We pray that, in the near future, thanks to the commitment of families and doctors, BTHS will not claim any more lives.

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