Quote of the Day:

"There is one word that comes to mind when I think of the Barth Syndrome Foundation and that word is HOPE. This group has given me hope for a future I once never dared to consider for my son. Now there is hope for the future when once there was none. *~ Allanna, Mother of Diagnosed Son, Scotland*

Featured Story: Alfie, age 12

My name is Allanna and my husband is Tommy, we have two sons, Jay and Alfie. Alfie was born in September 2000.

Alfie was a very sleepy baby and didn't seem interested in feeding and this, along with very odd bright green stools, was a concern for us. We spoke with our community midwife and after three days at home, Alfie was re-admitted into hospital. The doctors thought that Alfie's liver looked a "bit bright" and this, along with the fact that there was other family history of cardiac issues, made them transfer him to Yorkhill Children's Hospital two days later.

No one quite knew what was going on with Alfie. He had a brain scan, echo, metabolic testing and a liver scan, but doctors came to the conclusion that Alfie's heart was "a little big" but they were not worried and thought it was just down to his "bad start." We were sent home after four days. During the next few weeks, I switched him to formula milk just in case this was the problem but he was still a very quiet baby and not interested in feeding.

At his clinic in October 2000 he had another echo and we were told that he had dilated cardiomyopathy and he was admitted back into Yorkhill and started on Captopril and Propanolol. We were sent home after five days and were very frightened for our son. Alfie started to feed better and seemed to gain weight steadily for a while, but was never really interested when it came to starting solid foods; he seemed to prefer his milk over everything else that was offered to him. We were told not to worry about this as he was growing and seemed to be thriving on even the little amounts that he would take. Alfie reached his milestones a little late and we were told that this was probably because he had a brother 18 months older who was fetching things for him.

Alfie had numerous hospital admissions from around November 2002 with different "viral" bugs and about five episodes of ear infections. He twice had febrile convulsions with these infections. This was a horrible time for Alfie as he just seemed to get over one thing and something else would crop up. Barth syndrome was suggested around the start of 2003, through another member of my family. Tests showed that I was a carrier, Alfie had Barth syndrome, and Jay did not. We were really scared and I remember typing it into the computer and being absolutely terrified by what I saw!

We contacted the Barth Syndrome Trust and felt relieved that there was some positive news, support and advice on hand. We were added to the Barth Syndrome Foundation listserv (an e-mail forum) and found this service invaluable. If we had any question or concern about Alfie someone had some experience or advice to share or could point us in the right direction for the answers. We also found that the listserv archives were a great source of information. You won't find a more supportive bunch of people to share stories and knowledge about this rare genetic disorder than these. It feels like an extended family around the world who know what your son is going through and are on hand with support, understanding and genuine compassion.

We were then put in touch with Prof Kate Bushby at the Centre of Life in Newcastle who was wonderful! She started making sure that ALL of Alfie's symptoms were addressed, not just his cardiac care. Alfie started getting physio; he was put on prophylactic antibiotics to try and cut down infection; he was seen by podiatry; he had occupational therapy; and Prof Bushby requested that ALL Alfie's doctors copy one another into their letters so that his care was shared.

Alfie was tested over a four week period in May/June 2004 during which his neutrophil count wasn't higher than 0.3. Eventually he was put on G-CSF on the advice of Dr Steward in Bristol. Alfie seemed to thrive on G-CSF— no infections, no hospital admissions, and he is generally a livelier boy.

Alfie still does not have a great appetite and his diet consists of mainly milk, pasta, rice, noodles, cereal and other soft foods. He gets tired during the day but is learning to manage his Barth syndrome a little each day.

Alfie is currently in primary 5 at school, is enjoying good health right now (touch wood!) and living and loving his life! He is a very sociable boy with lots of friends and enjoys playing football with his brother, being out on his bike, and is part of a drama group. He also likes getting into mischief like every other child! My son has so much to battle against, but he is a wonderful boy and I am very proud and grateful every day to have such an amazing, beautiful, gutsy little guy in my life!

Allanna’s experience with her son and her warm and friendly personality are a great help to our families. Allanna and Tommy and their extended family and friends have also raised a considerable sum for BST over the years.