Quote of the Day:

"I am certain there are many families who share a family history like mine who have lost their young sons over several generations, totally unaware of the existence of Barth syndrome. The cause of those deaths remained a mystery until my son was diagnosed. While it is extremely stressful to be a parent of a very sick child, it is far worse to be a parent of a very sick child who doesn't have a diagnosis. A diagnosis of Barth syndrome has given me the ability to be an informed and empowered advocate for my son." ~ *Donna Strain, Mother of Diagnosed Son, Australia*

Featured Story:
A Personal Message from Donna

Donna and Cameron

Barth syndrome, although we didn't know, has a long family history with us. My knowledge of it starts back with my Nan. She had two girls and a boy and then lost three boys in a row. Failure to thrive they called it, and from what I can gather Nan felt very guilty about these little boys and she suffered depression and was never the same after. Aunty then had a little boy and he never grew properly. He died around six months of age and was barrel chested. He had poor heart function and my aunt had to crush up his medicine the best she could to give him. My mum then had a boy and by this stage she had an idea that there may be something familial going on. My brother, Martin, never went home. He fought for 10 weeks before he died and left my parents heartbroken. They told my parents after the autopsy that my brother had the heart of a 70 year old man and they called it fibroelastosis. My parents consulted a geneticist who advised them not to have any more children and to consider adoption. He told them that whatever this illness was it should have skipped a generation after my grandparents but obviously it hadn't. We know that this information is incorrect now, but my parents lived in the hope that if they managed to have children this disorder would not be carried by them. My parents took a gamble and had me and then my sister. We have since learned that we have distant family in the UK who also have children with Barth syndrome.

(L-R) Fraser and Cameron

In 1998 I fell pregnant with Cameron. It was a relatively normal pregnancy other than vomiting 24/7 for 24 weeks. Cameron was born two weeks early and was tiny at 5lb 12oz. Given our family history we saw a cardiologist and had an echo. Cameron's function was on the low side of normal and we made an appointment for another checkup in a couple of months. At nine weeks our world turned upside down.

Cameron was having trouble feeding and seemed snuffly. We went to our family doctor who promptly sent us to the hospital where we found Cameron was in heart failure. We were devastated. We were also very fortunate however as the cardiologist had recently read a paper on Barth syndrome, and given Cameron's presenting symptoms and my family history, he thought it very likely that this is what he had. Bloods were sent away and it was 18 months before we got a confirmation.
In the meantime, we consulted a geneticist who told us that with my family history we shouldn't expect him to live out of infancy and as you can imagine we were devastated.

We found the Barth Syndrome Foundation and with that we found instant friends, family, and acceptance. It can be a lonely world when nobody understands what you are going through and fighting for. We attended our first conference in 1996, and it was a very emotional and confronting experience, but it is one we have never regretted. We laughed, cried, and learned so much about Barth syndrome. When you are a parent of a child with a rare disorder you need to learn as much as you can as you are your child's best advocate. Doctors have lots of patients but we have only one Cameron and we are grateful for all of the research and hard work the Foundation has undertaken.

(L-R) Donna, Jordan & Cameron

Cameron is now 12 years old. He reached his milestones late but he reached them. His heart has been stable and he has only recently started having trouble with neutropenia. He has managed up until now to attend primary school full time but as the grades get higher he struggles more to keep up and the fatigue is greater. He uses a wheelchair for long distances and he is now finding that his low muscle tone is impacting on his friendships as he cannot keep up. He was recently told that he wasn't getting an invite to a birthday party as the child's Mother had said he couldn't be invited because of "his medical problem." It is so heartbreaking at times, you just want your son to fit in and be 'normal' and happy. Cameron loves the conferences so much because, as he says, “When you are there "people just get it." As much as Cameron hurts, he always manages a smile.

We were so lucky and blessed to get a diagnosis so early on and we are so lucky and blessed to have Cameron here with us today. He is a typical 12 year old, cheeky and a bit too smart for his own good. However, Barth syndrome has given him empathy and insight far greater than his years – but with that comes worry beyond his years also. Cameron loves life and lives it to the full. Most days he tells me, "I love my life mum." I just wish my Nan was here to see him.