By Mel, Mother of Affected Individual, Australia

Aiden (age 1)

In 1997, my entire family was genetically screened for Barth syndrome (BTHS). I was 19 years old, and results identified my grandmother, my mum and myself were carriers of BTHS. At the time, the results didn't have any effect on me; I was in 2nd year university, and having children was the LAST thing I was concerned about! The results did impact my parents though. They had lost their first-born son in 1970, my brother, Craig. He was three months, three weeks and three days old when he passed away, and the reason given was "heart complications." Conclusions have now been made that he suffered from Barth syndrome, and this was the reason his young life was taken.

What was most significant from the family's results was discovering that my Uncle Greg (mum's brother) had the BTHS gene. He was 45 years old at the time, and today is 61! The main symptom he shows is cardiomyopathy, and to this day he has never been hospitalised by illness. He is a true "Barth Wonder Story" and provides my husband Luke and me with real inspiration and hope for Aiden's future.

Because we knew I was a carrier of Barth syndrome, I had a Chorionic Villus Sampling (CVS) test performed at 12 weeks with both pregnancies. A CVS is a test that checks the pregnancy for genetic abnormalities. For our first pregnancy, the results came back "negative" for Barth syndrome, and gave us our eldest son, Sebastian. For our second pregnancy, the results came back "positive" for Barth syndrome, and gave us our second son, Aiden, who was born on May 26, 2011.

Due to his prenatal diagnosis of Barth syndrome, we had a full team of medical specialists attending Aiden's birth. As soon as he arrived, Aiden was taken straight to NICU, and apart from needing to be on continuous positive airway pressure (CPAP) resuscitation for two hours, his doctors were very happy with his condition. Initial echos showed an obvious spongiform thickening of the left ventricle with an ejection fraction (EF) of 26%, and contractility at the lower end of normal. He was started on Carvedilol on day five, and because of his better-than-expected condition, we got to take him home with us on day 10!

Life at home was fantastic! That is, until Aiden was three weeks old. It was a Tuesday, and I noticed that he wasn't feeding well at all, was quite breathless, and had a blue tinge around his mouth and between his eyebrows. We took him straight to emergency at the Melbourne Royal Children's Hospital, and he was admitted into the critical care unit in the cardiac ward later that evening, in severe heart failure. Our world crashed!

Aiden was in a terrible state, with his EF down to 16%. It was at this time we first met Dr. Andreas Pflaumer, Aiden's cardiologist. Dr. Pflaumer was then, and still is, someone who will remain in the highest regard within our entire family. Together with Aiden's other medical team, Dr. Lexie Frydenberg (pediatrician) and Dr. Joy Lee (metabolics), they have made an unbelievable effort in getting to know and understand the complex disorder and management of Barth syndrome.

During Aiden's stay in hospital, we first contacted the Barth Syndrome Foundation. The immediate welcome and support from the entire community was quite overwhelming. Living in Australia meant we were geographically disconnected from most, but the emotional connection we felt was very much instant. It was during these first few contacts when we were persuaded by everyone's recommendation to attend BSF's 2012 Conference.

(L-R) Sebastian, Mel, Aiden (age 1) and Luke

Attending the Conference was a huge learning curve for Luke and myself. While it was great to have individual discussions with Dr. Kelley, Dr. Steward, and the rest of the Barth medical specialists, the highlight for us was seeing and talking with all the Barth boys and men. Personally, my favourite memory was Amanda Clark taking the photo of all the "Barth Brothers" in the Grand Ballroom. Seeing them all standing together was a moment I'll never forget. On our return home, we felt empowered with knowledge about Barth syndrome and supported by all the life-long friendships that were made.

We feel very blessed to have Aiden in such a stable, mild condition at the moment. His latest cardio check showed that his heart function is within "normal" ranges, and his EF is now up to 61%! Even more exciting is he has just started walking at 16 months of age! Aiden is such a delightfully, happy young boy.

People are always commenting on his bright smiles and magnetic personality. Who knows what the future will bring, but by having the support of the Foundation and the Barth community behind us, we feel confident we can take this journey on together!