

Way to Go Sammy-Joe!

A lot has happened since Sammy-Joe's story appeared in the March 2003 NoticeBoard. In September 2003, with tremendous support from family, friends and the community, we were able to take Sammy-Joe to America to see some doctors who specialise in Trichothiodystrophy. Since the trip Sammy-Joe has improved out of sight—he even won a gold medal in his school Olympics!

My eldest son Sammy-Joe has been a mystery to doctors ever since he was born. After years of endless rounds of appointments and tests we finally got some answers. The doctors told us that Sammy-Joe has a rare condition called Trichothiodystrophy (TTD), for which there is no cure. This means that exposure to the sun or heat causes him intense pain. Sammy-Joe also has lactose intolerance, ataxia, speech delay, autistic characteristics, poor weight gain and stunted growth. Certain sounds and lights can cause him to scream as if in pain. He is also photosensitive (sensitive to light) and he often faints when he is unwell.

When Sammy-Joe was a baby, it was so difficult to convince doctors that something was wrong with him. Once we got the diagnosis things began to make sense—all those times I had told him to go outside and he had refused was because he was in pain.

I began to research all I could about TTD. Through the Internet I discovered that there were only 41 people in the world with TTD. There are 15 children with TTD in Australia, which has a high number of cases compared to the rest of the world. Sammy-Joe is the oldest survivor.

I was put in contact with two doctors in the United States (US) who had been researching TTD for over 20 years—Dr Cleaver at the University of California in San Francisco and Dr Kraemer at the National Institute of Health of Bethesda in Maryland.

They were particularly interested in Sammy-Joe because he was the oldest known survivor of TTD. They told us that sun exposure was also affecting Sammy-Joe's immune system and that he was just as sensitive to cold as to the sun.

The doctors offered to treat Sammy-Joe in the US if we were able to get him there. If we could raise enough money to get to the US they would cover our accommodation and all our medical expenses to treat him. But how could people like us raise thousands of dollars?



Sammy-Joe's story first appeared in the March 2003 edition of NoticeBoard.

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Maria with Sammy-Joe

We frantically set out to raise money so that I could get Sammy-Joe to America in September 2003. With help from a solicitor who kindly donated his time, we set up the Sammy-Joe Trust Fund and we started fundraising. The community was also very involved and helped to raise money through a dance held at Empire receptions and one held by Solarino Social Club. Community businesses donated items of food and products to be raffled at the dance and we held market stalls, all by donation of people in the community and my very good friends and family.

We were able to raise \$18,000 dollars and this covered the cost of flights, insurance and also some spending money. The hardest thing was having to leave my other son Christian and my husband Joe behind, but with lots of reassurance from family and friends that they would be looked after, we set off to America.

It was a hard three months but it proved very rewarding to Sammy-Joe's health. The treatment that Sammy-Joe received from the scientists and doctors in the US has had a big impact on his life and health. They helped me to understand his condition and they talked to me about the importance of protecting him from UV exposure and about how dangerous his illness can be.

They decided that if they could alleviate Sammy-Joe's anxiety and pain then he would have a chance to grow and develop. They believed Sammy-Joe was continuously

struggling to survive every day, as exposure to UV light broke down his DNA causing irreparable damage to his body and cells. The first step was to find out where his body was deficient and also to alleviate the pain he felt. Then they began Sammy-Joe on a course of vitamins and medication.

Although there is no cure for TTD, the quality of his life took a turn for the positive after this course of vitamins and medication. Sammy-Joe has now doubled his weight and has grown 16 centimetres since his trip. This is amazing because children with TTD usually need the help of growth hormone to grow. I had chosen not to give them to Sammy-Joe because I was worried about the side effects.

We also travelled within America also to see Dr Mark Pittelkow and Dr Digiovanna, who were also keen to examine Sammy-Joe's DNA. All the doctors are still in contact with us and keep us informed on all the latest news on TTD; even doctors in the Netherlands and France contacted us to examine his DNA.

Professor Rowe from the Children's Medical Research Institute (CMRI) in Sydney is keen to have research started here in Australia. He has told me several times that it is a miracle that Sammy-Joe is still alive as these children don't survive past a certain age. We agreed that we need to build up an alliance with the CMRI to have research started here in Australia. Genes for Jeans Day is an important event that happens every year to raise much needed funds for research on rare genetic illnesses. Once I got back from America I contacted them to tell them there were 14 other children in Australia with TTD. They too are keen to have research started in Australia.

I have set up a web site to tell Sammy-Joe's story and to raise awareness about TTD. With a group of other parents, I also began to lobby the government for more funding for genetic research into rare conditions. I have written letters to the Premier and my local MP and Sammy-Joe's case was even raised in Parliament.

Through the Starlight Foundation at the Royal Children's Hospital, Sammy-Joe's Starlight Wish was granted last year. His wish was to go to Queensland so we were able to go on our first family holiday—to the Gold Coast! Christian was so thrilled to have



Maria and Sammy-Joe with Dr Kraemer and Dr Di Giovanna from Bethesda Hospital in Maryland, US

a holiday by the beach. We chose very carefully the time of year when the UV in Queensland was at its lowest, which is in June. Sammy-Joe had to be covered up and protected from the sun but he still got to touch and play with the dolphins at Seaworld.

Sammy-Joe is amazing and he has changed so much since our trip to the US, he even won a Gold medal for running in his school Olympics. The race was held in the gym at school and he took his hats and jacket off and ran like the wind. Then he quickly put them back on, realising that the lights bothered him, but he didn't complain until after the race. He kissed the medal when he received it and the teachers were so proud of him.

I believe a mother knows her child better than anyone and that we have a basic and most accurate diagnostic tool at our disposal called A Mother's Intuition. If I had ignored that nagging feeling in my gut that I was right, that it had to do with the sun, I believe Sammy-Joe's condition would be much worse. Something kept telling me to trust my instinct and to pursue it further. I know I went to the extreme and travelled across the globe to be heard, but I have no regrets. At least now I know that Sammy-Joe was right all along and that I was right in all actions I took to be heard.

Maria Liistro

Sammy-Joe's web site is www.vp-it.com.au/sammyjoe

Yahoo TTD Support Group

<http://health.groups.yahoo.com/group/TTD-support>

MSN TTD Support Group

<http://groups.msn.com/Trichothiodystrophy-Australianworldwidesupportgroup>

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Sammy-Joe Trust Fund

ABN 34 585 645 433

Sammy-Joe's Trust fund is still running which goes towards the support group and another trip to the US so that Sammy-Joe can be reviewed in two years by the doctors in America. My next mission is to convince doctors, scientists and the government that we need to keep up with the rest of the world when it comes to starting research on rare illnesses such as Trichothiodystrophy, XP BS and Cockayne syndrome.

For more details or to make a donation contact Maria Liistro on 9305 6182 or Sam Barbagallo on 9390 9929 (Trustee).