

MEET OUR MEMBERS

Trichothiodystrophy (TTD) Support Network

"The TTD Support Network is a place for families as well as patients who are looking for information, contact and support for people living and dealing with trichothiodystrophy... We are not doctors or specialists, we are concerned parents who want to provide the best we can for our children"

About the TTD Support Network

The TTD Support Network brings together families of children with TTD from around the world. This group is founded by parents of children living with this disorder and would like to share and support other families living with the same. As well, it provides helpful information to those who provide care or interact with any persons facing this disorder.

The web site is hosted by Maria, whose son has TTD. It contains her story, and a link to her blog, where we get a glimpse of the day to day life of a child with TTD. The site also contains a factsheet on TTD, links to the online support group, media, and further reading on TTD research.

The TTD Network has a strong focus on raising awareness in the medical community

and among the general public. As a result, children with are now getting diagnosed a lot earlier, allowing parents to minimise their UV exposure.

History of the TTD Support Network

The TTD Support Group was started by Maria Liistro in August, 2002 after her son, Sammy-Joe, was diagnosed with TTD at the age of 13. Maria searched the net for answers, and found other parents after posting a plea on the internet and setting up a webpage. The group has been a huge success, and members feel like 'one big worldwide family' – one with 16 children in Australia and 57 other children scattered across the globe! The group stays in contact through an online messageboard, but hopes that one day face-to-face meetings will be possible.

About Trichothiodystrophy

Trichothiodystrophy is a rare recessive DNA repair disorder, in which the cells are constantly breaking down due to UV exposure and cannot be repaired. It is a seriously disabling disorder, with a severe skin affliction and serious developmental defects and growth retardation. Children with TTD are frequently sensitive to light and UV, and experience extreme pain and blistering, as well as fatigue, dizziness, headaches and other physical problems when exposed to sunlight.

Diagnosis

Diagnosis is made by studying the hair mounts, and by amino acid analysis which demonstrates decreased high sulphur matrix proteins. The hair breaks and fractures once it emerges from the skin and is exposed to the environment, and so the result is brittle, short and sparse hair.

Treatment

There is currently no treatment for Trichothiodystrophy.

Contact the TTD Support Network

Address: 13 Hursley Court, Craigieburn VIC 3064

Email: mliistro65@hotmail.com, sammyjo2@bigpond.com

Websites: <http://www.vp-it.com.au/sammyjoe/index.htm>

<http://www.caringbridge.org/ok/sammyjoe>

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

Trichothiodystrophy: a mother's story

My son Sammy-Joe has TTD and has only recently been diagnosed. He is now 17 years old. He was born by emergency caesarean section at 38 weeks. He was smaller than expected for a child at that age, and his head was 26% smaller compared to the norm. He was born with curly, brittle hair, short and sparse, small eyes, dry skin and very floppy. He weighed 5lb 2oz and lost a dramatic amount of weight in the first two days of his life.

He lost all of his hair after having a temperature, while having a bath in the hospital. He had difficulties suckling, and failure to thrive. Because of his poor immunity, he has had many stays in hospital throughout his life undergoing extensive tests, Gammaglobulin treatment, surgery and rehydration. He has lactose intolerance, ataxia, speech delay, autistic characteristics, poor weight gain and stunted growth. Certain sounds and lights affect him, triggering him to scream as if in pain, which I now believe at times truly hurts him.

In our world it is easy to understand things, but in Sammy-Joe's world it is challenging. His condition still challenges him in many ways, and gives him difficulties that other people probably wouldn't be faced with and wouldn't need to worry about every single day - things like worries about light, things like blisters constant head pain and severe burning feeling on his skin just from getting undressed to put his other clothes on. It has taught him tolerance, patience and most importantly understanding. I always say that regular people wake up and start a new day leaving all their worries from the days that have passed behind them - but not Sammy-Joe. He wakes up and his worries are still there and this gives him great difficulty in understanding why? He doesn't understand why he has to be sick, and says out loud "help me, the lights hurt me, the sun hurts me help me."

Aside from all the trials and battles in his life, he is a happy child that has a passion for books, videos, nursery rhymes, The Muppets, animals, stuffed toys and James Hird from the Bombers. My sister-in-law once said, "imagine if everyone had the same passion for things, or loved the same way Sammy-Joe does. This world would be a better place".

I have often been asked what does the future hold for my son's life, and I would be lying if I said it doesn't scare me. In fact it scares me so

much my response is always "nobody really knows". The other children with TTD that I am in contact with are all younger than him, and because this disorder is so rare, I have only been able to find 16 other children in Australia, 73 world-wide.

In 2003 we travelled to America to meet up with scientists and doctors from NIH and MAYO Clinic and UCSF Hospitals which helped change the quality of his life. Although not cured his quality has greatly changed.

Sammy-Joe is my pride and joy, my heart and soul, and my whole reason for living. He is truly a miracle child and a survivor, an inspiration to his family and friends. His brother Christian adores him, and he is the apple of his Dad's eye. We love both our children and have taught them to be very proud and stand tall, despite all the challenges that life brings us.

Sammy-Joe constantly amazes me with his incredible happiness and his sheer love of life. Like all parents, we want everything for our son, and thinking ahead about what the future may, or may not hold, makes me strengthen my determination to see that my child has every opportunity and experience he deserves. Sammy-Joe's diagnosis has forced us to re-evaluate our entire lives and the way we live. We surround ourselves with family and friends who understand and are sensitive to our situation. We decided as a family that whilst this diagnosis is devastating and we have no control over it and the final outcome, we do have control over the way we manage our situation and the way we choose to give Sammy-Joe a quality of life that best suits him. The simple things in life really do mean much more, and spending quality time with Sammy-Joe means so much more to all of us. We all live day by day and enjoy every moment with our precious little boy.

I'd like to tell you all that as long as we are blessed with Sammy-Joe, we will have all that we need. Everything else seems so unimportant. He will always touch hearts, and even change lives with his smile.

More of Sammy-Joe's story can be found on the TTD website. Thanks, Maria and Sammy-Joe, for sharing your story.