PROFILE

Dylan Finglas

LIVING WITH MULTIPLE SULFATASE DEFICIENCY (MSD)

"Dylan started to gradually regress after 3 years of age, losing all gained skills."

Dylan Finglas, is 7 years old and was diagnosed with Multiple Sulfatase Deficiency (MSD) just after his 2nd birthday in late 2014. The diagnosis was very quick after a private consultation at Temple Street Hospital. Some of Dylan's best achieved skills were walking with assistance, being able to throw a ball with a reasonable amount of accuracy and being able to feed himself a biscuit.

Dylan started to gradually regress after 3 years of age, losing all gained skills. He lost the ability to walk and eventually to sit up unassisted. Any words he had were lost as was the ability to feed orally (now peg fed) and his eyesight has deteriorated to the point that he is now blind. Although Dylan is mentally and physically disabled now, he loves music and will laugh and smile most days with stimulation. He absolutely loves hydrotherapy and this is greatly beneficial as he is non active. His family are saddened by the lack of hydrotherapy facilities that exist.

The early diagnosis, especially given that Dylan was so well at diagnosis led his family to establish the first ever charity to advocate for MSD research among other rare disease patient advocacy. They are called MSD Action Foundation/SavingDylan.com.

RARE DISEASE

About Multiple Sulfatase Deficiency (MSD)

Multiple Sulfatase Deficiency, an ultra-rare condition, is a clinically devastating and fatal condition in children. Depending on age of onset, children affected by MSD, in most cases, do not live long enough to see their 10th birthday. The condition is neurodegenerative and progressive. It is classified as a lysosomal storage disorder.

The deficient gene is called the SUMF1 gene. Mutations or deletions on the gene result in a misfolded controlling enzyme called the Formylglycine Generating Enzyme. As a result, substrates build up in cells in the body including the brain and they become toxic. The myelin sheath does not form properly in the brain as a result. MSD is similar to Metachromatic Leukodystrophy and Sanfilippo Syndrome.