PROFILE

Liam and Saoirse Heffernan

BATTEN DISEASE

"Children become totally disabled and eventually lose all bodily functions"

Two Children who were featured in the original Easyguide (2013) were Liam and Saoirse Heffernan who both had the ultra rare condition known as Batten Disease. Saoirse Heffernan died in 2011 aged 5 and Liam, her brother, died in 2014 also aged 5. The lives of their two children inspired their parents Tony and Mary Heffernan to establish the well known and respected patient advocacy group 'Bee for Battens'. In tribute to their memory and to the work of Bee for Battens, The Saoirse Foundation and BUMBLEance, we reprint the profile that was in the 2013 Easyguide.

Liam has an ultra-rare condition known as Batten disease, which is an inherited genetic disorder of the nervous system that usually manifests itself in childhood. Batten disease is named after the British paediatrician who first described it in 1903. It is one of a group of disorders called neuronal ceroid lipofuscinoses (or NCLs).

Early symptoms of Batten disease (or NCL) usually appear in childhood when parents or doctors may notice a child begin to develop vision problems or seizures. In some cases, the early signs are subtle, taking the form of personality and behaviour changes, delayed speech, slow learning, and clumsiness or stumbling. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills. Children become totally disabled and eventually lose all bodily functions. In Liam's case, he was diagnosed when he was just 18 months old, following the diagnosis of his sister, Saoirse. Batten disease is not, at this time, preventable. To date it has always been fatal. Saoirse passed away on Jan 18th 2011, aged 5 years, 7 months and 14 days.

On May 3rd 2011, Liam became the youngest ever child to undergo brain surgery as part of a treatment trial at Weill Cornell Hospital in New York. While the immediate months and first year after the procedure Liam showed significant signs of improvement, unfortunately, Liam has now started to deteriorate, and his parents face the fact that they will lose their remaining child to a rare disease.

RARE DISEASE

About Batten Disease

Batten's disease is an ultra rare, inherited disorder of the nervous system that usually manifests itself in childhood. It is one of a group of disorders called neuronal ceroid lipofuscinoses (NCLs). While it affects only a tiny number of children in Ireland, it is believed to be severely undiagnosed. The forms of NCL are classified by age of onset have the same basic cause, progression and outcome, but are all genetically different. The defective gene, inherited from both parents, causes malfunction at a cellular level. Early symptoms of Batten disease (or NCL) usually appear in childhood with vision problems or seizures. In some cases the early signs are subtle, taking the form of personality and behaviour changes, delayed speech, slow learning, clumsiness or stumbling. Autistic traits and dementia can also feature quite severely. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills. Children become totally blind and disabled. To date it has always been fatal. Being an ultra rare condition, international collaboration in research is proving most successful in attempts to learn more and find a cure.