We felt so lost and scared until we found the ED support group UK

When Harley was one, we took him to see doctors multiple times as his nails were peeling off, his teeth were starting to decay as quickly as they came through and he was suffering from a lot of pain, episodes of which we now know are seizures. We just knew something wasn't right, they told us he was playing on us, to give him calpol and a hug and that was it.

Eventually, nearly three years on, I did a post on Facebook and a friend recommended I try talking to a lovely pharmacist she knew.

Reaching a Diagnosis

I emailed the pharmacist with a description of everything that was happening, along with videos and photos, who finally listened and agreed with me that there was something not right. He had a friend that was a doctor, who he forwarded our emails over to and he agreed to refer us to the hospital. The paediatrician at the hospital then in turn referred us to a specialist hospital, who diagnosed him with Ectodermal Dysplasia, Shapiro syndrome and recommended Harley have an MRI due to the symptoms he was showing.

Six weeks later, he was put under general anaesthetic, now aged four, and had the MRI. Six short days later we received the news that Harley had grey matter heterotopia (PVNH) in multiple parts of his brain.

We were absolutely devastated and completely thrown into the unknown. I asked him what did this mean? And he just passed us a Google page!

We felt so scared and lost until we came across the Ectodermal Dysplasia support group in the UK and the PVNH Support group page on Facebook.

They explained everything so much better, and so much of it now made sense.

This was just over a year now, and Harley has had many hospital trips and stays since then, along with many more tests.

He has also since been diagnosed with epilepsy and has been put onto seizure medication, although he still gets very tired it has been amazing. He has gone from having six+ seizures a day to about three a month. We are hopeful that it continues to help.

We are still awaiting genetic tests (trio genome sequencing test, epilepsy panel and cerebral malformation panel) and some results of some other tests, but we feel we are finally getting somewhere.

Finding support and treatment for Harley

Harley is now under five different hospitals and seven different consultants. He has recently been referred to rheumatology due to problems he is having with his joints. He cannot walk for long periods of time and needs additional support from an SEN buggy. He struggles to hold pens due to pain he suffers with in his hands.

Harley has sinus tachycardia, hyperhidrosis, problems with temperature regulation, a low immune system (a simple cold can hospitalise him), suspected ADHD and learning developmental delay.

He has missed so much due to his conditions, including a lot of time off school. But in September last year, he started back part time with a one to one and is absolutely loving it.

Harley makes us so proud every single day and he takes everything in his stride. He has been through so much in such a small period of his life, but most of the time he faces it all with a smile on his face.

"I really cannot thank all the team at the ED Society enough!

They have helped me so much. From supporting me through Harley's DLA application forms, to giving training to his school to help with temperature regulation, other symptoms, what needs to be done, and just general support through some of the hardest times.

Along with all the other group members in the ED community that understand in one way or another, I really don't know how we would've gotten through the last year without their kindness, love and support."