HAEUK

Rachel's story

Living with HAE

My name is Rachel and I was born in 1978. At the time, my family were unaware that HAE existed within our family.

When I was about 18 months old my parents began to notice that I was being sick on a regular basis, but the doctors were not convinced there was anything abnormally wrong with me. Throughout my early school years I was regularly sick and suffered abdominal cramps, at least once or twice a fortnight, and on the odd occasion I had swelling in my hands or feet.

Over the years I was tested for various food allergies and spent months on special food diets, but none of these seemed to make a difference. My problems were classed by doctors as migrane tummy, for which I received no treatment.

Although this iterruped my every day life, thankfully I generally managed to miss no more than one day of school at a time due to an attack, so I didn't fall behind too much with my schoolwork. It was difficult though, as friends and teachers didn't understand, and I had no reason to explain why I had severe cramps and felt sick, tired, feint and unable to concentrate at times.

In 1993, aged 15, I attended yet another appointment with a specialist, who finally decided to test for HAE. He said it was extremely unlikely this what what I had, but he was otherwise was at a loss as to what was wrong with me. The tests came back positive, and finally I had a diagnosis.

Because of my diagnosis, we now managed to trace it back through the family and realised this was the cause of my fathers and grandmothers abdominal attacks, although rare, and my late great grandmothers facial swellings, the cause of which had always been a mystery.

After my diagnosis I started attending regular outpatients appointments at Southmead hospital, with an immunologist who helped me put together a treatment plan to manage my symptoms. The medication I began taking was Danazol, and for the next 17 years, combined with Tranexamic Acid, I remained fairly healthy with just a few attacks. This came at a great time, helping me through my GCSE exams and college years, and enabling me to secure a full time job.

My symptoms over the years have changed frequently. In my earlier years I suffered with mostly abdominal cramps, sickness and diahorrea, and in my teens I started having regular attacks of oedema, mainly in my hands and feet. More recently I often develop a rash before swellings appear, and the swellings are generally in my hands, feet, knees, elbows, hips and groin area, the latter of which making it extremely uncomfortable when sitting or walking around. The abdominal cramps and sickness are also regular symptoms, and these seem to develop much more rapidly then when I was younger. Thankfully I have only ever had a couple attacks of odema affecting my throat and airway, all of which are frightening and reminds me just how important it is to have a emergency supply of C1 INH at home.

Because of the prolonged use of Danazol, I had regular blood tests to monitor my C1 levels as well as ultrasound scans on my liver area. Over time I developed a small adanoma on my liver which caused me a little discomfort, and was monitored closely with regular MRi and ultrasound scans.

Due to personal reasons, in 2010, in consultation with my immunologist, I decided to take a break from the Danazol completely. Within a few weeks of ceasing Danazol, the tenderness in my liver area started to subside, and a scan a few months later showed the legion had shrunk in size.

Unfortunately, two weeks after stopping the Danazol I suffered a very bad night of abdominal cramps and sickness, and at 4am awoke with severe jaw ache. Over the course of a few hours this spread to my neck, shoulders and then arms, and after attending A&E at 8am, I was later diagnosed with suffering a minor heart attack. This came as a huge shock to me, as at the age of 32, I was fit, playing sport a few times a week, ate healthily and was a non smoker. The cause of the heart attack has never been diagnosed.

I began having regular infusions of C1 INH when needed, which was every 5-6 days, but this meant regular trips to my local hospital for it to be administered. This made planning my every day life difficult because my attacks were so frequent and developed rapidly, that I was afraid to be too far away from the hospital.

At the time, funding for home therapy was held by local primary care trusts and were dependant on individual funding requests. I applied for funding to self treat at home but was refused twice. Many months later, and on my second appeal, I was finally granted the funding and was taught to self infuse. This was such a life changing time; I finally had the freedom and confidence to travel away from home and could plan a holiday with my family.

Although HAE does have a big impact on my day to day activities, I am determined to not let is stop me doing the things I enjoy and leading a full and active life.