HAEUK

Ann's story

Living with HAE

The story of hereditary angioedema (HAE) in our family is a very positive one. Although it started with much ignorance, fear, pain and mismanagement, we now understand the disorder and have confidence and enough control over the situation to live full and active lives. We hope that in the future, as new therapies come onto the market, the prospects will be still better. I have told our family story to encourage those who have had to struggle, and perhaps continue to struggle, with many years of misdiagnosis and inadequate treatment.

Let me start by introducing our family as we are today. I am a 67-year retired nurse. My husband John, also 67, is a health and safety specialist. We have three grown up children: two sons and a daughter.

My own symptoms first occurred when I started taking the contraceptive pill at the age of 23. I immediately experienced severe bouts of gross oedema about once a week, involving arms, legs, face and gut. It was very frightening. Only at this stage was I told the family history of this weird condition. My grandfather had HAE severely over many years and he died from a throat swelling at the age of 39. My father suffered severely until he was 30, when his symptoms suddenly ceased, never to return. Several aunts and uncles were affected. My mother regarded the condition as "all down to nerves" and "all in the mind". My doctor prescribed massive doses of antihistamines to no effect.

My symptoms continued, but gradually decreased over the next few years. Doctors were emphatic that there was no connection with the pill, but when I discontinued it the oedema stopped completely. When I became pregnant with my first child I had regular oedema from four months, but all symptoms ceased after her birth. The same pattern occurred during my second and third pregnancies, although the symptoms were more severe. However, between the pregnancies I was free of symptoms.

Our third son appeared to be a very healthy baby but our lives were devastated when he tragically died in infancy.

It was at this time that my GP spent an afternoon researching HAE at the postgraduate medical library on our behalf. Subsequent blood tests revealed that my children were, like me, deficient in a substance in our blood called C1-inhibitor (C1INH). However, they were symptom-free at the time. Because of these diagnoses I was determined to find some answers. As a nurse I had access to medical literature and our GPs were supremely helpful. We found a specialist consultant in London. Gradually over the next few years more information and treatment became available.

Two years after the loss of our second son we had another baby boy who also has HAE.

In 1983, towards the end of my last pregnancy, I was the first patient in this country to have C1INH injections. It was like a miracle. What had been a three-day session of very acute abdominal pain, vomiting

and diarrhoea was relieved within an hour of the injection.
The availability of C1INH has been the central factor in transforming the lives of our family. Without treatment, for instance, my daughter would not have been able to manage a career and she would have experienced a lot of severe pain and distress. Her symptoms had increased in severity since the age of 14. She suffered severe incapacitating abdominal symptoms for three days out of every seven to ten days. At first she was given high doses of Danazol, but the androgenic properties of this were prohibitive. Her symptoms are now controlled by self-injecting two vials of C1INH intravenously as soon as abdominal or throat symptoms develop. Thanks to this replacement therapy my daughter has been able to qualify as a doctor. She now lives a normal and happy life and has travelled all over the world. She has three children of her own - two of them have been diagnosed with HAE.

My oldest son developed problems in adolescence, but his condition has been well managed with low doses of danazol and home possession of C1 Inhibitor to inject in the event of attacks of angioedema. He always keeps C1INH available in case of severe symptoms or laryngeal emergencies. Because of such a good HAE management regime he has had six years at university and has travelled round the world.

My youngest son is now 28 years old. He manages his symptoms very well by self injecting C1 Inhibitor in the case of an attack, and occasionally using danazol at times of particular risk. With full approval and wise advice from his Consultant, our son climbed Mt Kilimanjaro this summer!

Our five year old grandson has not had any HAE symptoms yet. Sadly our six year old granddaughter has had severe attacks every week to ten days since she was three years old. However she has home therapy with C1 Inhibitor early on in each attack, and she enjoys a very happy and full life.

We know how very fortunate our family had been over the years. We are under the care of a brilliant Immunologist who has set up individual HAE management programmes for each of us. We all have a home therapy program that has enabled us to keep safe and have a very good quality of life. None of us has ever needed to go to A&E, and none of us has had to take days off work or school because of HAE.

Sadly over recent years many patients have struggled to get the right diagnosis. Many people have suffered for years without a referral to a specialist with experience of HAE management, and because of the financial constraints of the NHS there are many patients who cannot get funding for treatment that will enable them to have good control over their HAE symptoms.

HAE is a very rare condition and we need a patient association so that we can work together to help all HAE patients achieve good control over their symptoms and enjoy a good quality of life.