

Andy's story

Living with HAE



Family History

HAE came to me through my mother's side of the family. Mum was very sick all of her life and passed away in 1969 at the age of 24. Her death was caused by a swelling in her throat, unfortunately medical attention came too late. I was just five years old at the time and I then went to live with my paternal grandparents.

Unfortunately my mother's death caused something of a rift between my mother's family and my father's family so I didn't see much of my maternal family during my childhood. My grandfather was apparently a sufferer too, although he lived well into his seventies. Both he and my mum found it difficult to be taken seriously by doctors at the time. My grandfather was branded as 'workshy' and my mother was actually treated in a mental health hospital for some time.

Swellings In Childhood

As a child growing up I would often get swellings in my hands, feet or face. These were usually brought on by a bump or a fall and the swellings would be incredibly huge, but not at all painful. I was always told that it was 'oedema' and stuff like 'Oh, your mum used to get that' but it was never explained to me properly. I don't think anybody really understood it back then. The doctor would give me some tablets to reduce the swelling and it would go down after a couple of days, so I never really worried about it.

Early Adulthood

As I grew into adolescence the swellings seemed to slow down. At the age of eighteen I broke my ankle and there was another huge swelling at that time, but after that it seemed to stop altogether.

When I got married in 1987 my wife, Jackie, and I discussed the condition and decided to find out more about it. Before we had any children we went for genetic counselling to find out the risks of passing on the condition to any children. We were told that the risks were basically 50/50 and as I was in very good health we decided to go ahead with our family.

My son, Joel, was born in 1990 and after a year we were able to test him for the condition and I'm very pleased to say that he came up clear.

Stomach Problems

In the early '90s I began to suffer from occasional chronic stomach upsets. They were excruciatingly painful and would begin with constipation and sickness, leading on to diarrhoea towards the end of the episode. At first the doctors said it was a recurrent virus, but the episodes began to get more frequent and more serious, causing me a lot of discomfort and a lot of time off work.

I began a series of tests at the local hospital, I had every test imaginable. They looked for ulcers, colitis, Crohn's disease, cancer and anything else you can think of. All the tests kept coming up negative so eventually they decided I had Irritable Bowel Syndrome (IBS). They began to treat me for IBS but the treatment was ineffective and my health was deteriorating.

Eventually the attacks were happening approximately every two weeks! Life was becoming increasingly difficult for us as a family. Joel was still very small and couldn't understand what was happening. Thankfully Jackie was extremely supportive through all the hard times that were to come, without that support life would have become impossible.

Hospitalisation and Diagnosis

In August of 1994 I woke in the middle of the night and was throwing up (as usual!). This time it was different as the vomit contained some blood. We called a doctor and he had me hospitalised. At last they began to take it seriously.

I was in hospital for about a month. For the first week I was on a drip and not allowed to eat anything. Lots more tests were carried out. Then one day the doctor came to see me and said that the only thing they could find wrong with me was that I had a very low level of C1 Esterase Inhibitor.

Of course I knew that I had HAE but I had just never associated it with this kind of illness before, As far as I knew it just caused my hands and feet to swell. What we discovered was that the HAE was causing my intestines to swell and that when that happened the tubes of the intestine would close up, effectively stopping the process of digestion. Hence I would get constipated and vomit because the food couldn't pass through my system. The pain of these attacks was so bad that I almost wanted to give up and die. As the attack wore off and the swellings went down, so I would get diarrhoea as my system cleaned itself out.

My father was able to tell us that my mother used to get similar attacks before her death. It would have been helpful if he could have told us that a bit earlier but now that we all knew what we were dealing with it was up to the doctors to decide how best to treat me.

The case was not helped by the fact that whilst in hospital I contracted pneumonia and got very sick indeed. My weight dipped to seven and a half stone and I came pretty close to death at one point.

Treatment and Side Effects

The first treatment the doctors came up with involved an injection of Fresh Frozen Plasma (FFP) at the onset of an attack. This would introduce a higher level of C1 Esterase Inhibitor (C1 INH) into the bloodstream and help to bring the attack to an earlier end. This was initially quite effective, although it took some time for the treatment to work. They then discovered that they could obtain Concentrated injections of C1 INH which had been pooled from donated blood. These were very expensive and every time they gave me these injections they insisted on telling me how much they cost. Thank God for the National Health service! These injections worked much faster than the FFP and I was soon able to go home. Over the next few months I was still getting attacks quite frequently but I was at least able to go straight to the hospital and have these injections, I could then return home within a couple of hours.

The next problem to address was to find a way to stop the attacks happening.

One thing we discovered was that the attacks seemed to be brought on by eating certain types of food. All I

can tell you is that I know there are certain foods that will put me in hospital. These include very spicy foods like curry or chili, any fried or greasy food (including chips), red meat or fatty meat and a few other things. These days I eat a ridiculously healthy diet, mostly vegetarian and including lots of rice and pasta, trial and error has led me to a diet I can safely eat.

I had suggested to the doctors that they try giving me the C1 INH on a regular basis, but I was told this would not work as it would not supplement my blood properly. Their suggestion was a steroid called Stanozolol, which would help the blood to produce a higher level of C1 INH.

Initially I was put on 2.5 mg a day and this was quite effective for a while. After a couple of months I began to notice some side effects which included freaky mood swings, headaches and very greasy skin. After talking to the doctor about this the dose was reduced to 2.5 mg every other day and the side effects lessened, although I still suffer from very severe headaches quite regularly.

The attacks were now less frequent but were still occurring approximately every three months.

Introduction of Tranexamic Acid

My case was transferred to the University Hospital in Cardiff and the immunologist there suggested the use of Tranexamic Acid in combination with Stanozolol. Initially I was put on a dose of 1000 mg twice a day. Over the following months we juggled with the doses and I am now taking 1500 mg twice a day and 2 mg of Stanozolol two to three times a week

Current Good Health

I've been following this drug regime and diet for a few years now and in general I am in very good health at the moment. I am always aware that every HAE sufferer's case is different and that it affects people differently at different stages in their life. Hence I am always keeping an eye on myself. Although, see the latest update below for some news!

Recent Swellings

These days I still have occasional episodes of swelling in my hands , (like this one) so I am aware that I may be beginning a new cycle of episodes. In August of 2004 I had a major abdominal attack that really caught me by surprise, it was the first attack like this for eight years. Until 2003 I had never met another sufferer, outside of my family, and all my family members who have suffered from HAE are now dead, so I have felt very alone in this. It's good to be able to talk to other sufferers and compare notes with people from all over the world.

Throat Swelling

My HAE is pretty well under control these days and I rarely get attacks so this one caught me on the hop a bit back in 2008.

I woke up with a sore throat and I guess that probably started things off. By lunchtime I was finding that I couldn't swallow and could only manage a bowl of soup. I wanted to cough but wasn't able to and the back of my throat felt inflamed. I wasn't struggling to breathe at this point, but it was getting worse. I left home for work at 2.30, but by the time I got there I had decided that it was time to go to the hospital, because breathing was now becoming a bit laboured. I was a train driver at the time and I couldn't take a chance on

things happening once I was in charge of a train. So I told the manager when I got there that I needed to go to the hospital and one of the managers took me there.

Fortunately I had my own supply of C1 INH with me. I always carry it with me when I go to work in case of an attack when I am too far from home and that has now proved to be the right thing to do!

Despite having the C1 and a letter from my immunologist there was still a bit of fuss about giving me the injections, but taking my own supply definitely sped things up and after about half an hour the C1 was being administered.

The A & E doctor was trying to tell me that I should have an epipen and that when she examined me she couldn't see any swelling in the throat. But then my immunologist and immunology nurse showed up, as we were in the same hospital at Cardiff where I am registered. When he examined my throat the immunologist said he could see a definite swelling and that I had obviously done the right thing.

After about an hour the swelling had started to settle down. The C1 infusion did the trick which tells me that this was indeed a HAE episode and not just an inflamed throat infection.

This was quite a scary episode for me and even more so for the misses! I had never had a throat swelling in my life before, so I wasn't sure what it was going to feel like. I've always advised people not to take a chance with throat swellings and I'm glad that I took my own advice. If I'd got on that train this could be a very different story today. My mum died from a throat swelling back in 1969 so it's always something that is in the back of my mind.

Since that time my HAE has settled back down again and I have had no major swellings, but you never know what the next day will bring.

REMEMBER : DON'T TAKE A CHANCE WITH A THROAT SWELLING!