

Our Story



A Mom and an Advocate

By Lynsey Jayes

I'm currently 30-years-old and living in Leeds in England, although I'm originally from Aberdeen where most of my family still lives. As a child I remember my Granda for two things - always wearing a cap and always having cotton wool up his nose.

I remember Granda started getting really ill when I was about 10, and around 1994 is when I first remember visiting him in the hospital, where he seemed to spend most of his time in and out. He was eventually diagnosed with hereditary hemorrhagic telangiectasia, which he taught us how to say as kids.

I first started getting little random bleeds around 19 when I had my first son, although nothing major. I remember my cousin being bothered with nosebleeds when he was playing football in his early teens. And since Granda dying, it became apparent that three of my uncles and my mum had started being bothered by nosebleeds and red spots in the same way that Granda had.

My first born son, who is now 11, started getting nosebleeds around 3-years-old. I took him to the doctors a couple of times and was always told that it was probably an infection of the little vessels. Gradually, the nosebleeds got more frequent, but now it's just something he has learned to get on with. He sorts them out in school or gets up and sorts his nose out during the night without bothering anyone. He's just used to it because he hasn't known any different.

Then my youngest son, Michael, was born in 2012 and started getting nosebleeds at only a few months old. These got worse as he neared his first birthday, and he would bleed most times he cried. He is now just turned three and gets random bleeds at all times of the day.

I took him to the general practitioner, and as my nosebleeds had gotten worse by then, and I'd recently been told I had Ulcerative Colitis due to intestinal bleeding, I started to piece everything together. I wondered if this HHT Grandpa had was actually the culprit, so I mentioned it to the doctor when I went with Michael.

The doctor's reaction to my asking if Jacob, Michael and I actually could have this HHT was, "That's not something that shows symptoms until middle age. How old was your mum and your grandad, etc.?"

However, I wasn't convinced so I went back again with Michael and Jacob after having found information on the HHT Centre at Hull and demanded the three of us be referred there. The doctor just sort of shrugged me off and said he would get his secretary to look into it.

I chased the surgery up for months afterwards and eventually got referred and seen in April 2013 at Hull by Prof Morice and Dr. Robinson at the HHT clinic. They explained that they wouldn't expose children to radiation unless they displayed symptoms of PAVMs, but that they would all be called for screening when they reached puberty. My sons were diagnosed based on telangiectasias on the face, nosebleeds and familial history.



They did, however, screen me since I had been to my general practitioner on various occasions for breathlessness. They did a CT scan on me and low-and-behold there was a large AVM with two feeding vessels on the left lower lobe. I had that coiled off in June and was advised to tell all my family members who suffer with HHT to be screened as none of us had ever heard of PAVMs before.



I will now be screened annually for any further AVMs and the kids will start PAVM screening in a few years. We have since found out that we have the Alk1 gene because of my mum seeing the geneticist. She has had many tests including a positive echo bubble, but a year on and we are still waiting for a simple CT to locate any AVMs.

It's very frustrating to see how difficult it is to be heard, treated and screened for anything HHT related in the UK. I am very lucky to have managed to get my referral straight to Hull as it seems many doctors are reluctant to do this. The lack of understanding is quite scary, and the fact that I was told my kids can't have HHT until they are middle-aged by a trained medical professional is quite worrying.

But being armed with the information from the net and demanding that my doctor refer me could have potentially saved my life. I'd been walking around with a large PAVM that hadn't been picked up on chest x-rays for more than two years.