Our Story

Denial and Acceptance of HHT

By Tami Lyvers

In 2003, at the age of 29, I didn't feel quite right so I went to the doctor to have my thyroid tested. I have thyroid disease, so I thought my levels were probably a little off. I told my doctor my symptoms, and then remembered I had written them down on a post-it note. I had forgotten to add short of breath - that's how slight it was. I didn't even remember to tell the doctor without looking at my list.

She took my oxygen with the oximeter, and it was 88 percent. She had me get an x-ray, saw a spot and treated me for walking pneumonia. I returned in a week for my check-up, but showed no improvement, so she recommended a CT scan. I was resistant because of the cost and the hassle. I asked if maybe my oxygen was naturally low (I didn't know anything about oxygen levels at that time). She said no, you really need to get the CT done. I agreed and went in right away for a CT.

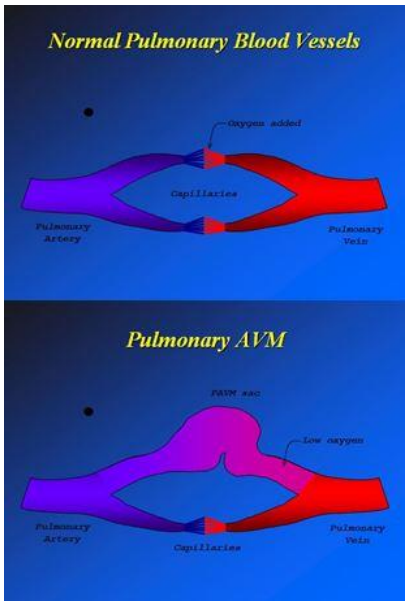
Afterward they decided to admit me to the hospital and put me on oxygen. My levels didn't change, of course. After a while a nurse came running in and said with urgency, "Get that IV out of her arm." It was really scary because she was clearly worried, and I had no idea why. The other nurse rolled her eyes and took her sweet time removing the IV. She was annoyed at being told what to do and clearly didn't understand what the big deal was.

My doctor came in and told me I had a pulmonary arteriovenous malformation in my lung. She gave me a basic explanation with a page out of a medical textbook with one very small section on PAVMs. She told me it was extremely rare, and they would have to find a specialist to treat it. The excerpt from the text said one in every 50,000 people has PAVMs. One of the nurses was so excited to learn about a new rare disorder he said he would research it once he got home. To me it wasn't exciting - It was terrifying.

I tried to research it on the internet, but there was very little information I could find. I felt alone and scared. I remember discovering HHT online and feeling isolated because I couldn't find anyone like me who had a PAVM without HHT. My family and I felt my AVM was a fluke. I just had the one (and haven't had any since). I rarely have nosebleeds, and I don't have the characteristic telangiectasias. We haven't had any terrible tragedies as a family, such as people dying young because of stroke

or abscess. We ignored the fact that both my mom and sister have fairly frequent nosebleeds. We felt that most people have nosebleeds at some point in their lives. Nosebleeds are common, we reasoned.

I had a coil embolization in March of 2003 to fix my AVM, and I haven't had any problems since. Then, in 2012, during a routine colonoscopy, doctors found that my mom's oxygen was very low. She immediately told the doctor about my AVM, and during the colonoscopy they found AVMs in her colon. Her physician then ordered a CT scan right away and sent her to a lung specialist.



The lung doctor recognized she had AVMs from the CT scan, but wanted to do all sorts of other tests to make sure something else wasn't causing her low oxygen. We all knew this was absolute nonsense, and that she needed to get in with the doctor who had treated my AVMs right away. As it turns out, they got her in the day after viewing the scans because my mom had four PAVMs, one of which was very large. She was 64-years-old at the time. We are so lucky she didn't have some terrible consequence because of our previous denial of HHT or the various doctors' lack of knowledge.

My sister was also tested, and the doctors found she had a lung AVM as well. So far, all of our PAVMs have been successfully treated, and our MRIs have not shown any brain AVMs. As a family, we have been very lucky that we have not had the tragic consequences of the disease so many others have. We pray that we continue to remain healthy.

I read others' stories and my heart goes out to the families who have experienced such tremendous loss because of this disease. We have been so fortunate that tragedy didn't strike when the rest of my family was not tested when my AVM was first discovered. But it could have gone the other way.

No doctor suggested having my family tested for HHT. It was not even mentioned. I believe the medical professionals thought, like me, that my AVM was a fluke. Or maybe they had never heard of HHT. If doctors become more knowledgeable about this disease, it will save lives. I was on my own in my research and learning about HHT. And I did almost all of that research after we found out my mom had a lung AVM. That could have been too late.

I had a great doctor with a wonderful ability to fix AVMs, but without knowledge of the disease that often accompanies them. My mom and sister could have found out years earlier about their AVMs. They had been needlessly walking around for nearly 10 years with these ticking time bombs in their lungs, and I am so grateful that nothing bad happened.

We have since had genetic testing done on the whole family. My son was diagnosed with HHT as was one of my sister's three sons. My recommendation to family members of people with HHT is to absolutely get tested whether you have symptoms or not. I thought of myself as someone who did not fit the criteria of HHT - My nosebleeds are extremely infrequent and minor, I have no telangiectasias and we had no catastrophic family history indicating this disease was present. But I do indeed have an AVM and HHT. If you have a first degree relative with an AVM and/or HHT, get tested. It could save your life.

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