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Young Mom Haley Miller Battles Rare Disease, Von Hippel-Lindau, at U of M

By Rick Marshall

Awful diseases can have funny names; or, put differently, some afflictions can be obscure because of confusing descriptions. Everybody knows what Parkinson’s Disease is, but not who Parkinson was (by the way, he was a British geologist and paleontologist). This writer’s wife suffered from Lewy Body Syndrome, named for the German doctor Friedrich Heinrich Lewy, who isolated and investigated the protein that causes a certain form of dementia. Describing the disease’s name was a constant component of answering my friends’ questions, until the comedian Robin Williams’ suicide, which was partly ascribed to Lewy Body Syndrome.
This article will be about a brave woman who battles one of those rare afflictions, a gene mutation with a strange name. Most of us seldom think about how devastating some diseases can be. … until it hits close to home.

The rare Von Hippel-Lindau (VHL) disease was diagnosed during tests on Haley Miller when she was 20. She underwent genetic testing, then a relatively new science, and was diagnosed with VHL. She has been under treatment since then, but possible changes in healthcare policies might compromise Haley’s ongoing treatment. But she is a fighter, and so is her medical team, and so are various support groups. She is being monitored by specialists at the University of Michigan, principally the noted Endocrinology Team.

According to the VHL Alliance, an advocacy and educational organization, VHL is a genetic condition characterized by tumors in up to 10 areas of the body. These tumors can be benign (harmless) or malignant (spreads to other parts of the body). Most of these tumors are benign, but VHL tumors in the kidney and pancreas can grow to a stage where they become malignant and can spread to other parts of the body.

While tumors or cysts in the spinal cord and brain are benign, they can be serious. As they grow in size, these tumors can cause an increased pressure on the body structure around them. Because it is impossible to predict how the disease will present or progress, regular surveillance is extremely important for people living with VHL. Those affected by VHL undergo annual body scans to check for progression and changes in the disorder. Haley Miller began those regular examinations, and much more, including having family members tested. “It was around 1999,” she recalled, “… and the doctor recommended our whole family go, so we did.”

Most families don’t just think of setting up genetic testing, according to a U-M profile of Haley, but when her father was diagnosed and eventually passed away from complications of Von Hippel-Lindau, the Miller family knew it could only help.

“There was a 50-50 chance I would have it. My father was a twin and both he and his twin were the first mutations in our family.” The results came back positive for VHL for Miller and her sister; but her brother’s results came back negative. After hearing the results, Miller arranged for a scanning protocol.

“The weird thing was we had already set up the scans, and then about three weeks before they were scheduled to take place, I started having symptoms similar with the disorder. Before that I had felt fine,” she says. “They decided to bring me in early and my symptoms just seemed to get worse.”

The VHL surveillance guidelines were developed to make sure that VHL tumors do not cause additional damage to the body. With careful monitoring, early detection, and appropriate treatment, the most harmful consequences of this gene mutation can be greatly reduced, or in some cases, completely prevented.

Haley, now a wife and mother of two, mentioned that she sees many specialists while she was pregnant both times, including having extra testing, to make sure her condition is kept under control. "I worked with the doctors and specialists while I was pregnant both times, including having extra testing, to make sure everything was going smoothly," Else says. "As a comprehensive care center for VHL, we work very closely with an excellent network of experts at U-M covering all potentially affected body parts. For example, if we find a tumor in a patient’s brain during his/her scans, we send the patient to an excellent neurosurgeon for further treatment.”

Haley mentioned that she sees many specialists and her protocol is to watch and monitor her body. This was especially important when she and her husband decided to expand their family. "I worked with the doctors and specialists while I was pregnant both times, including having extra testing, to make sure everything was progressing normally for me and my babies,” Miller told the Healthblog. “And once each of my daughters was born, I worked with the genetics clinic to also have them tested. My oldest daughter is fine, but my youngest daughter has acquired the condition, too. She will come with me to the genetics clinic every year for medical care.”
Haley points out that research is an important part of the fight against VHL. “I want people to know you have to donate and fund research to find a cure for this disease. Many researchers believe if they can find a cure for VHL, they think they can find a cure for cancer in the future.”

Dr. Else said that it is patients like Haley Miller who are willing to donate their tumors to a research registry that helps researchers find out more information about the condition.

“Almost 100 per cent of patients consent to being part of the tumor registry because they’re interested in learning more, and know it can potentially help find future treatments,” he said. “Research for VHL is really only possible because registries like this exist.”

Even though Haley currently lives with benign hemangioblastomas (tumors in her central nervous system) and several cysts in her pancreas and kidneys, among other locations, she considers herself lucky.

“Both my sister and I have VHL, but luckily we haven’t had to have several surgeries. This disorder can be very difficult to live with and monitor for most people. Even my father and uncle had several surgeries by my age.

According to the VHL Alliance, people with VHL disease are at risk for developing:

• Hemangioblastomas, benign tumors on the spine, brain, or retina
• Kidney cysts, benign kidney tumors, or clear cell renal cell carcinoma (kidney cancer)
• Pancreatic cysts or pancreatic cancer
• Pheochromocytomas or paragangliomas, tumors of the adrenal glands
• Cystadenomas, benign tumors in the reproductive tract, affecting the epididymis in men and the broad ligament of the uterus in women
• Endolymphatic sac tumors, benign tumors in the inner ear
• Benign lung lesions
• Liver cysts

Some people with VHL disease might experience tumors in multiple areas, but in some cases people might only be affected in one area. Because of the uncertainty of exactly how VHL will manifest, regular monitoring and surveillance (see Sidebar) is very important to effectively manage the disease.

Regular check-ups and surveillance are important for anyone living with a chronic disease. However, it is even more important for people living with VHL disease, given the unpredictability of the condition. Early detection, regular surveillance and appropriate treatment can greatly reduce the most harmful consequences of this gene mutation.

It is important to diagnose and begin screening children who are at risk as early as possible. Using DNA testing, it is possible to determine if a child has a VHL mutation and needs to follow the recommended screening.

Haley Miller strives to manage her condition these days, and doesn’t let it control her life, she told the U-M Healthblog.

“In day-to-day life, it doesn’t affect me much. I try to live a pretty normal life and be diligent about my scans every year, and keep ahead of it.”
VHL disease is different in every patient, even in the same family. Since it is impossible to predict exactly how and when the disease will present for each person, it is incredibly important to continue to check for all the possibilities throughout a person’s lifetime.

People who have VHL disease may experience tumors and or cysts in up to ten parts of the body, including the brain, eyes, kidney, and adrenal glands.

**HOW DO PEOPLE GET VHL?**

VHL disease is a hereditary condition, meaning that it can be passed from parents to their children. The disease is caused by a change, or mutation, to a gene referred to as VHL. If either parent has the VHL mutation, there is a 50% chance they will pass it on to their child.

People who do not have VHL disease have two normal copies of the VHL gene, one inherited from the mother and one inherited from the father. People who have VHL disease are born with one working copy and one altered copy. This alteration, called a VHL mutation, destabilizes the second copy of the gene allowing tumors or cysts to grow.

Most people with VHL inherit a gene mutation from a parent. However, approximately 20% of people with VHL have a new VHL mutation not passed down from a parent. The alteration occurred before birth. In these instances, although there is no prior incidence of VHL, the person will still have a 50% chance of passing it on to their children.

VHL has a high penetrance rate, meaning that most people with VHL will experience one or more tumors at some point in their lives. The penetrance rate for VHL is 97% by age 60 years according to researchers.

**IS TESTING IMPORTANT?**

DNA testing is the best way to determine if someone has VHL. This is done through a blood test and must be processed at a clinical testing laboratory. If you or a family member has a family history, or believe you may have VHL disease, talk to your doctor or genetic counselor about DNA testing. Early diagnosis can help you manage VHL effectively.

**SURVEILLANCE**

The VHL Alliance and its Clinical Advisory Council recommend screening children should begin as early as age 1. Early screening includes an eye/retina examination, a hearing test, blood pressure measurement and a neurological assessment.

Beginning at age 5, imaging tests such as MRI or ultrasound are added to aid in early detection and management of any VHL tumors. It is important to detect VHL tumors before any symptoms occur in order to prevent or minimize any adverse effects.

Participation in VHLA’s Cancer in Our Genes International Patient (CGIP) Databank allows participants to generate screening reminders.

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### FAQS ABOUT VHL

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