**MAY IS VHL AWARENESS MONTH!**

**By Barbara Correll, Vice Chair, VHLA Board of Directors**

Von Hippel-Lindau (VHL) Awareness Month, held in May each year, presents a great opportunity for providers, patients, caregivers, and the community to raise awareness about VHL. Raising awareness improves diagnosis and treatment, increases fundraising opportunities, and ultimately will help contribute to finding a cure.

Most people—and even some healthcare providers—have never heard of VHL, so increasing awareness is vitally important.

Greater awareness leads to early detection, diagnosis, and improved management of the disease. Like any disease, when people know what they are facing and are armed with the information they need, they are better able to take the appropriate action to manage their health. At the same time, greater awareness can help uncover new funding sources and research opportunities to help uncover a cure for VHL.

What can you do to participate in VHL Awareness Month? How can you raise awareness?

- **Educate people you know.** Talk to your friends, family, co-workers, or other groups about VHL. Share information about VHL. Tell them your story or about someone you know with VHL.

- **Explain the connection to cancer.** For people who do not know about VHL, it may be hard for them to understand. Explaining the connection to other cancers creates relevancy for people who do not have experience with the disease. The following is a simple way to explain this connection:

  - **In healthy people,** the von Hippel-Lindau (VHL) gene controls cell behavior and prevents tumor formation. In VHL disease, the gene does not function properly, causing tumors to grow in up to 10 different parts of the body. Research on von Hippel-Lindau and greater understanding of the disease will lead to a cure for many forms of cancer.

- **Share stories on social media.** Share your story—or your friend’s or loved one’s story—on Facebook, Twitter, Instagram, or other social media outlets. Stories about real people make VHL relatable and can help people better understand its impact.

- **Include photos in your post**—they help the post get noticed and put a “face” to it.

- **Organize a fundraising event.** VHL Awareness Month is the perfect time to host a fundraising event. Not only is it a good opportunity to raise funds, but you can also raise awareness about VHL. Need ideas? Check out vhl.org/get-involved/fundraise or connect with Heidi at Heidi.Leone@vhl.org.

- **Be an advocate.** Talk to healthcare providers and other stakeholders in your community. Share information about VHL and VHLA. The VHLA website (vhl.org) is packed with great information and resources you can share.

- **Volunteer with the VHLA.** If you’ve ever thought about volunteering with VHLA, VHL Awareness Month is a great time to start. Remember VHL awareness does not stop in May! There are a variety of opportunities and needs, ranging from social media support to writing to contacting your local news outlets or sending a letter/email to your friends and colleagues. For more information, contact info@vhl.org.

- **Get involved and help raise awareness today!**
The VHL Patient Databank:

PROVIDING A MORE COMPLETE STORY OF VHL

The various manifestations of VHL are well known in the VHL community of patients, caregivers, healthcare professionals, and researchers. We know that lesions can develop in up to 10 different parts of the body: retina, kidney, spine, brain, adrenals, pancreas, inner ear, reproductive organs, liver, and lungs. But how else does VHL impact a person physically and emotionally?

Thus enters the VHL Patient Databank, a natural history study designed to further advance the understanding of VHL. Data collected is essential if we hope to improve diagnosis, treatment, patient outcome, and ultimately find a cure.

How many VHL patients have wondered about their thyroid issues or digestive complaints? How many have experienced persistent headaches or panic attacks? Have you ever noticed that your mouth is dry? Rest assured you are not alone. The presence of these issues surpasses levels reported in the general public. While we do not yet know what these medical findings mean, we know that they exist. AND we learned this through your data!

Now that we know many VHL patients experience these problems, it is important that the healthcare community addresses them, including incorporating these issues into patient care. Clinical studies are also needed if and how these ailments can be prevented and treated.

The VHL Patient Registry is the first study of its kind, using data entered by patients themselves. This novel approach allows information to be collected that may not be part of other studies, including clinician-driven natural history studies. In this way, the Patient Databank compliments and supplements the work of VHL researchers. The international aspect of this Patient Registry also has many advantages, including the fact that it contains data from all the genetic variants of VHL. As such, a comprehensive picture can be determined while differences between DNA variants can be compared.

The VHL Alliance just launched significant updates to the VHL Patient Databank, and we urge all patients to participate (vhl.org/databank). It is your data that will create the full story of VHL. Remember, your data might hold that “missing link” to finding a cure!

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VHLA-SPONSORED RESEARCH

THE VISION PROJECT

By Raymond Kim, MD, PhD, FRCPC, FACMG, VISION Principal Investigator, Internal Medicine and Medical Genetics, Assistant Professor, Department of Medicine, University of Toronto, Division of Medical Oncology

The complex, rare nature of VHL and its multiple manifestations creates unique challenges for patients, clinicians, and researchers. The best way to understand VHL is by collecting the experiences of clinicians and patients across a wide geographic area. Currently, VHL patients receive care and have their genetic testing performed in many different clinics and laboratories across the globe. This contributes to having separate, isolated clinical and genetic data of VHL patients, which makes it challenging to assess the information about VHL as a whole. This ultimately hampers the care of VHL patients.

To overcome this difficulty, an efficient process to share patient and genetic VHL data is needed. With the support of the VHL Alliance, our research group has initiated the Von Hippel-Lindau data and Information technology Sharing International cOnsortium (VISION).

One challenge in VHL research is limited information about the clinical relevance of thyroid issues, digestive complaints, persistent headaches, panic attacks, and continued on next page
identified VHL gene variants. Since testing of VHL variants is performed in different laboratories around the world, one laboratory may be unaware of VHL variants and the clinical interpretations that another laboratory has already identified. Sharing this type of data is essential to further improve our knowledge base about VHL. This problem also highlights the fact that unlike other hereditary disorders, VHL has not benefited from a collaborative, international effort to understand the VHL genetics.

Enter VISIon. The ultimate goal of VISIon is to share VHL genetic and patient data between the teams caring for VHL patients around the world, maximizing the benefits of the collective knowledge. Our research group is currently utilizing existing genomics information databases to address this problem: the Washington University School of Medicine’s CIVIC and the National Institutes of Health’s ClinGen and ClinVar. The CIVIC project will aid in sharing interpretations of clinical relevance of VHL gene variants. The database arm of ClinGen (ClinVar) will house our VHL gene variant and clinical patient data in a freely-accessible format. **All data will be de-identified!** We are working to make ClinVar the most up-to-date and comprehensive database to facilitate the dissemination of VHL information worldwide.

In order to accomplish this goal, we are reviewing all published VHL studies and existing VHL databases. This information will be used to update the CIVIC and ClinVar databases, as well as the comprehensive collection of VHL patient and variant data previously published by our research group in 2013. This effort will include more recent studies in underrepresented regions such as Ireland, Sweden, Korea, India, China, and Brazil. These data will augment the existing knowledge and help to propel the understanding of VHL. In addition, we will supplement this with information from the clinical experiences of major care centers in Toronto and the Netherlands.

We hope that in uploading our re-annotated data into ClinVar and CIVIC, we will be establishing a workflow that can ease the process of collaboration with researchers worldwide. Inviting other researchers to join the data collection effort is crucial to creating and maintaining the most comprehensive and inclusive genetic VHL database. Being able to relate genetic information with clinical experience may help with tailoring disease predictions for individual families and modify surveillance protocols.

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**AWARENESS**

**TAking ACTION!**

Written by Cari P, patient, caregiver, and advocate

My name is Cari. I am from Ohio. My 13-year-old daughter Alyssa and I both have VHL. Back in 2001 when I was first diagnosed with VHL, I held several fundraisers for the VHL Alliance, to help raise awareness and support research to find a cure for this disease. Now in 2017, I am taking action again. My mother died in 1982 due to an undiagnosed VHL brain tumor. Her doctors treated her as if she was crazy; they did not listen to her complaints of headaches and dizziness. Her experience inspired me to write a book called “All in Her Head,” where I share our story. Writing this book has been a wonderful experience for me, and now many opportunities are coming my way. I am becoming a public speaker, which is providing me the opportunity to share my story with many people—on the radio as well as at in person events. An independent film director has expressed interest in turning my book into a movie!

"I encourage you to take action, too."

I believe that the best thing that has come out of this experience is raising awareness of VHL. One by one, I am educating people about VHL!

I encourage you to take action, too. It could be through your own book or a public speaking event. For example, I am sharing my story with several local middle and high schools. It is amazing because I never thought my story would inspire so many people. Your story is meant to be shared and to inspire as well. We all have strength and greatness within us. Keep moving forward. Spread awareness. Raise money. We need to be the voice for our children who deserve to have so much more hope for their future. “All in Her Head,” is available on amazon.com, and I am donating a portion of the proceeds from my book to the VHL Alliance. Thank you for your support!
The VHL Alliance is pleased to recognize Massachusetts General Hospital as the newest Comprehensive Clinical Care Center (CCCC). Under the leadership of Dr. Othon Ilioupoulos, MGH serves over 200 VHL patients on an annual basis.

This has also been a busy season for renewing Clinical Care Centers: 6 CCCs and 6 CCCCs were renewed in April! The renewal process occurs every two years. Each center must submit an application which is reviewed by members of the VHL Alliance Clinical Advisory Council, many of whom are internationally renowned VHL specialists. In response to CCCs’ requests for more patient-centered feedback, we also sought feedback from VHL patients/caregivers who receive care at these centers. We are very encouraged by the commitment of these institutions to providing outstanding holistic, coordinated care for VHL patients.

The following CCCs received renewals: University of Alabama at Birmingham, University of Kansas, University of Miami, Mayo Clinic (Rochester), Washington University (Siteman), and Lahey. And the following CCCCs received renewals: Cleveland Clinic, Columbia, MD Anderson, Stanford, University of North Carolina, and Yale.

Please consider sharing your feedback about the CCC at which you receive care by completing this survey: bit.ly/HowWasYourVHLvisit

The VHL Alliance and each CCC values your feedback!

“*If you build it, they will come*”

This statement has never been more true than in our recent development of a VHL Clinical Care Center that meets VHLa’s standards. As a facility that serves much of the Intermountain West (Utah, Idaho, Wyoming, Montana, and parts of Nevada and Arizona), our patients are used to traveling quite far and, until recently, the nearest CCC was many states away. While we served many VHL patients in the past, becoming a CCC has highlighted the expertise we have here at the Huntsman Cancer Institute to be best suited to provide integrated and comprehensive care to our VHL patients. Recently, a young woman with VHL called and asked if she had the right number because she had von Hippel-Lindau. When I told her she had called the right number for our VHL clinic, she immediately started crying of relief that she had found a place that not only knew what VHL was, but had the background to provide her surveillance care. Here at Huntsman, we feel as if the benefits are mutual—not only do patients get more local care for VHL, but our multidisciplinary team gets to learn alongside each other at VHL tumor boards and research conversations.”

Samantha Greenberg
CCC Point of Contact, Huntsman Cancer Institute

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**Clinical Study Recruitment**

*Announcing a new Phase 2 clinical study for patients with: 1 or more measurable ccRCC tumors with no tumor greater than 3.0 cm and has a VHL diagnosis based on a germline VHL alteration.*

The National Institute of Health (NIH) is currently recruiting patients with more sites likely to open in the future.

If interested, contact Martha Nonos, RN: 301-435-8897; mninos@mail.nih.gov.

Principal Investigator: Ramaprasad Srinivasan, MD
Study Sponsor: Peloton Therapeutics, Inc.

Dear VHL Urologist, Nephrologists, and Oncologists,
I am in my 30s and lab results show I have low testosterone. My primary care provider is willing to prescribe testosterone supplementation. Could this testosterone supplementation increase VHL tumor growth? What things should I discuss with my primary care provider before deciding if this is a good option for his low testosterone?

—Proactive Warrior

Dear Proactive Warrior,
There is a lack of data regarding the effect of testosterone and VHL tumor manifestation, mainly because testosterone is not tested or replaced in young people unless there are obvious abnormalities. (Note: Androgen receptor expression has been reported in ~20–30% of sporadic renal cell carcinomas. Its role is still unclear with opposite reports.)

Having said that, we appreciate your question and encourage you to continue being VERY CAUTIOUS in replacing testosterone. Anabolic hormones have been linked to higher incidence of cancer. Body builders that take anabolic hormones, for example, have been reported to have a higher incidence of cancers in young age. Also, HYPOthyroidism has been associated with better prognosis in renal cancers.

To suggest proper treatment, your primary care provider would need more information, specifically: HOW LOW is “low testosterone” and WHY does it needs to be replaced? These points were particularly emphasized because of the fact that the patient is young. Problems of muscle mass, erectile dysfunction, or hair loss can be treated symptomatically rather than with testosterone administration. For example, ask your doctor if clomiphene may be appropriate.

If your primary care provider does determine that replacing testosterone is the best option for you, you are strongly encouraged to follow tumors very closely with higher imaging frequency for the first years to ensure that it is not increasing the rate of tumor growth.

Finally, please share this in the VHL patient databank! The more information that patients share about non-traditional treatments they are using the better we can answer questions like this. If a non-traditional treatment works, we want to help spread the news! If it is harmful for VHL patients, we also want to share the news so other VHLers can avoid unnecessary suffering.

Sign up and return to the databank annually at: vhl.org/databank.

BUILDING ALLIES FOR MY VHL JOURNEY

by Lauren S, VHL Patient

As a 25-year-old with VHL, I have been fortunate enough to find an amazing team of health care providers at the University of Chicago to take care of my medical needs. When I was diagnosed in 2014, I already had been living with Crohn's Disease for twelve years, so I had a leg up on communicating with medical professionals about my health needs. However, VHL is a completely different disease, with other considerations and timelines, and I was initially overwhelmed.

Luckily, I found the VHL Alliance and got in touch with my local chapter leader, Ellen. It was so helpful to talk to someone who had some experience living with VHL and to get another younger person's perspective. I attended my first Annual Family Weekend and got involved in local fundraising initiatives. Both were a great way to meet other people with VHL. They also helped me broach the subject of the disease with my family and friends, and get them involved in fundraising for a cure.

I am currently participating in VHIA's pilot Health Wellness Coaching Training program, which is an incredible learning experience. The training will allow me to work together with my local Comprehensive Clinical Care Center and help support other VHL patients and loved ones as they manage all of the complex emotions that surround living with a chronic disease.

As a young adult, there are many challenges that can arise because of VHL, including: family planning, facing multiple surgeries over one's lifetime, and managing the costs of health care. Through VHIA, I have met many people who are living with VHL and are generous enough to share their stories, and who are committed to working to finding a cure. The various support groups, the newsletters with research updates, and the events have all helped me connect with professionals and patients alike to not only have a better understanding of the disease, but to also build a community that will support me as I journey through life and manage my VHL.
In any ongoing medical or emotional condition, the caregiver plays a crucial role in the wellbeing of patients. That caregiver could be a spouse, child, or close friend.

In 2013, the biopharmaceutical company Shire published its Rare Disease Impact Report. Their survey research in the US and UK revealed that caregivers of rare disease patients felt feelings of depression, stress, isolation from friends/families, less interaction with friends/family, worry based on future outlook of disease, and anxiety based on lack of information available on disease. These feelings were higher in the situation where treatment options are limited, such as the case for VHL where there is no pharmacological treatment and patients and their loved ones face multiple surgeries.

Shire’s findings were not a big surprise to the VHL community. In 2011, a group in The Netherlands published data demonstrating that approximately 25% of partners of VHL patients exhibited clinical levels of distress that warrant psychological intervention.¹

The VHL Alliance is all too aware of the needs of the caregiver as they juggle many jobs in addition to taking care of their loved one. Because VHLA believes that caregiver burnout needs to be prevented at all costs, we have created a number of programmatic opportunities and resources specifically geared for caregivers. These include:

• The Caregiver Center on our website: vhl.org/patients/caregiver-center
• Telephone support groups facilitated by a VHL Caregiver: bit.ly/VHLSupport
• Staff monitored, private online community: facebook.com/groups/vhlcaregivers
• Better Together Peer Mentoring: bit.ly/VHLSupport
• Face-to-face meetings at our Annual Family Meeting: vhl.org/tampa2017

Please contact the VHL Alliance (info@vhl.org or 617.277.5667 x4) to get more information and to sign up for any of these programs. We hope that you will participate in one or many.


Support groups have been demonstrated to relieve anxiety, depression, and stress.

—Channing Paller
VHL MEDICAL MEETING IN ARGENTINA

On March 2-3, 2017, a multidisciplinary VHL Medical Meeting was held on at the Hospital Italiano of Buenos Aires in Argentina. About 120 health care professionals, students, patients, and families attended.

Presentations included clinical and surgical aspects of VHL, management of disabilities (e.g. rehabilitation of vision and audiology), genetic counseling, family planning, psychosocial aspects, and epidemiology of genetic mutations and research in Argentina.

Among the physicians and researchers presenting were Dr. Lucas Tedesco, a researcher at CONICET, who discussed advances in research (also presented at the 2016 International VHL Symposium in Boston, MA). Javier de Arteaga, a nephrologist from Córdoba, discussed his experience treating people with VHL.

The meeting was extremely valuable to patients and families as well as medical professionals in Argentina.

YOUNG ADULT RETREAT

June 9-11, Nashville, TN

Come to sunny Tennessee for the weekend! Share your experience as a VHL patient, increase your knowledge of caring for your holistic needs, and discuss topics that are particularly relevant to young adults. This weekend retreat offers a good balance between fun activities and serious discussions.

More details available at: vhl.org/about/news-events/young-adult-retreat/

DETAILS:

Young adult VHL patients (19–34 years). No partners or children. (Wait to bring them to the family meeting in Tampa this October!)

Cost: $50 registration fee + money for transportation and dinners.

Lodging: A home has been donated just south of Nashville for the retreat. Sleeping bag/air mattress/ couch space is available for Friday and Saturday night for the first 10 people who register.

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2017 VHLA ANNUAL FAMILY WEEKEND

OCTOBER 13-15, 2017 — TAMPA, FLORIDA

Fundraising Dinner & Auction
Annual Family Meeting
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2017 Team VHL 5K Run/Walk

Tickets and Registration: vhl.org/vhlweekend2017
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