Everyone at the VHL Alliance would like to extend a big Thank You to all of the patients, families, caregivers, researchers, healthcare professionals, and donors who supported us in 2016. Our work would not be possible without the tremendous level of support we receive. Our goal is to increase the quality of life and to find a cure for people living with von Hippel-Lindau. We will do it through research, education, and awareness. Nearly ninety percent of our funds are spent on our education and research programs.

VHL does not give up its secrets easily. The healthy VHL gene controls cell behavior and prevents tumor formation. In von Hippel-Lindau disease, the gene does not function properly, causing tumors to grow in up to 10 parts of the body. Research on von Hippel-Lindau will lead to a cure for many forms of cancer.

We are so lucky we are dealing with VHL today as opposed to a time in the past. There has never been more promise for the prevention and treatment of this disease. Some of the best and brightest minds in the world are working on the VHL problem. For example, people like Eli Wallace who we met at the VHL Medical Symposium in Boston last year. He works for Peloton Therapeutics. It’s a small company in Dallas, TX that has a big interest in VHL. He told us about an exciting new drug they are developing which targets Hypoxia Inducible Factor-2α (HIF-2α). That is one of the key players involved in oxygen sensing at the cellular level that causes big problems for people with VHL. The drug is in a Phase I clinical trial right now. And, based on data from our Patient Databank, they will be working with the VHL Alliance for patient recruitment and developing endpoints for a more expansive clinical trial. Bottom line—our data speeds up the trial and gets us closer to a cure.

Our Patient Databank is starting to reveal valuable information about the “natural history” of von Hippel-Lindau. It is so valuable that it has caught the attention of the National Organization for Rare Disorders (NORD) and the Food and Drug Administration (FDA). At NORD’s Rare Disease Summit, FDA Commissioner Margaret Hamburg hailed the Databank as a tool that can improve the agency’s ability to review treatments for rare diseases. Big data holds big promise for finding answers to VHL. Improvement means a faster review and a faster time to market. So I urge you to sign up for the Patient Databank at VHL.org/databank. If you have already signed up, please return! This is a long-term project. Your data might hold that “missing link”.

2017 shows great promise for advances in the understanding of von Hippel-Lindau. Solving this VHL problem takes time and money. And it also takes commitment by patients, families, caregivers, researchers, doctors, and donors. Winning this fight will always be a team effort. Let me assure you that all VHLA’s stakeholders (the patients, families, caregivers, researchers, medical professionals, and donors) are “all in” for the battle to help us cure this disease.
We all dream of a cure for VHL. Finding this cure will take hours and hours of painstaking research. And researchers need data—lots and lots of data. This requires participation from ALL of us.

Our family is new to the world of VHL. It gave us great hope to learn that there is a databank collecting information about VHL patients. The Cancer in our Genes International Patient Databank is an international partnership between patients and clinical researchers and was developed at the recommendation of VHLA’s Research Council.

Initially diagnosed and treated in Europe, our child now lives in the US. There is no question in our minds about the importance of sharing medical records from both countries. Teams developing clinical trials for VHL patients have already contacted VHLA expressing interest in using the patient databank. This research may save the lives of VHL patients. Yet I was stunned to learn that not many people have signed up to participate in the databank, and even fewer people are returning to participate longitudinally. The more information that is submitted, the greater the chance researchers have in finding answers about VHL.

Submitting your medical records is an important part of the databank, and VHLA makes it very simple: download a consent form from the website, complete it on your computer with an e-signature, and send it to VHLA—all in a matter of minutes. The VHLA will then request the records and enter them in the database. All data is stored safely using the same security technology used by hospitals and research facilities.

Each one of us is a unique link to solving the frustrating riddle of VHL. Participating in the database is a simple step you can take to contribute to improving treatment and care, and perhaps someday finding a cure.
Adrenal Issues & Surgery

Masha Livhits, MD, Endocrine Surgeon at UCLA School of Medicine discussed VHL in the adrenal gland. Pheochromocytomas (pheos) occur inside the adrenal gland and paragangliomas occur outside. VHL patients are predisposed to develop these tumors which are often non-cancerous but produce too much adrenaline, the “fight or flight” hormone. Symptoms can be violent (panic attack), less obvious (elevated blood pressure), or non-existent.

Asymptomatic adrenal tumors are fairly common for VHL patients, so it is especially important to follow the screening recommendations. For example, under anesthesia for an unrelated surgery, a “hidden” (undiagnosed/asymptomatic) adrenal tumor can create a life-threatening spike in blood-pressure. Patients who need anesthesia and know they have an adrenal tumor should receive pre-operative medicines (α-blockers and sometimes β-blockers) to help prevent this crisis. Patients who do not know if they have a pheo should be tested before any scheduled surgery. Emergency surgeries without the proper pre-operative medicines should be avoided. Screening for VHL patients can include a blood test, 24-hour urine test, and various types of scans.

Once a patient is diagnosed with a pheo or a paraganglioma, it should be surgically removed even if it is asymptomatic. Again, pre-operative medications which are taken for 2–3 weeks are vital! α-blockers help control blood pressure. They can also make patients tired and thirsty, so patients should make sure to stay well hydrated. β-blockers are sometimes added if a patient’s heart rate needs to be lowered.

In the 1950s, open adrenalectomies were considered the gold standard. In the 1990s, laparoscopic (entering through small incisions in the front) and retroperitoneal approaches (entering through small incisions in the back, with fewer surrounding organs to manipulate) became available, dramatically decreasing the healing time. Open adrenalectomies, however, are still used if the surgeon is concerned about malignancy or if the pheo is too large. The techniques preserve some of the adrenal cortex so patients can hopefully avoid taking steroids for the rest of their lives. Although this is promising, there is an increased risk of recurrence because the only way to ensure that all the tumor tissue is taken out to remove the whole adrenal gland.

Retinal Lesion Detection and Treatment

Michael G. Gorin, MD, PhD, Professor in Ophthalmology and Chief of the Division of Retinal Disorders and Ophthalmic Genetics at UCLA School of Medicine, discussed retinal lesions. Long-term surveillance is critical because up to 70% of VHL patients will have retinal angiomas, and the age of onset ranges from age 3 to 50+. Unless the retinal angioma is on the optic nerve, Dr Gorin suggests “if you can see it, treat it.” There is no good strategy for observing retinal lesions away from the optic nerve. Early intervention preserves vision by preventing the angiomas from growing to a size where they leak behind the retina and cause a retinal detachment. Larger lesions are definitely more difficult to treat. Here is a summary of treatment options which are available:

External cryotherapy is a painful treatment in which tumors are frozen in an uncontrolled way by applying a probe to the outside of the globe.

continued on next page
Endo-cryotherapy/intraoperative cryotherapy is more controlled and has better outcomes, but it requires the probe to be applied directly to the tumors, for example, during a vitrectomy operation.

Radiation is generally not a good option because VHL patients have a long time to live during which the radiation treatment can cause ongoing damage to their retina.

Traditional argon laser is relatively ineffective because the laser gets reflected off of larger tumors instead of penetrating/killing the tumor tissue.

Photodynamic therapy uses a special type of dye and an infrared laser to increase the uptake of laser into the tumor. While promising in theory, clinical outcomes are still variable and the dye is very expensive.

Anti-VEGF therapy requires injections in the eye. While it has had some success in reducing tumor leakage, it does not kill or shrink the tumor.

Fluorescein-potentiated argon laser (FPAL), Dr. Gorin’s preferred method, requires the use of a blue-light laser. After giving a patient fluorescein dye, the angioma will “light up like a light-bulb” and absorb blue light, which enables the ophthalmologist to destroy the tumor with little damage to the surrounding tissue.

Visually Impaired Phone Discussion Group
Interested in being part of a telephone discussion group for those with low or no vision? Contact office@vhl.org or 617.277.5667 x4.

CCC CORNER

VHL Alliance welcomes three new Clinical Care Centers (CCCs):

- Johns Hopkins Hospital [Baltimore, MD]
- Indiana University, Simon Cancer Center [Indianapolis, IN]
- University of Utah, Huntsman Cancer Institute [Salt Lake City, UT]

These CCCs join the ranks of over 50 centers worldwide recognized for providing outstanding coordinated care for patients with VHL. Check out the map of CCC locations at: vhl.org/ccc.

In addition to having specialists that are experienced in caring for patients with VHL, each CCC has a Lead Team of 2-3 people serving as the Sponsoring Clinician, Point of Contact, and VHL Patient Navigator. The Lead Team works together to ensure patients do not miss important scans or opportunities for participating in research projects or clinical trials. They also make sure patients’ emotional health is being monitored. As we all know, living with VHL can be a rollercoaster, and we want to do everything we can to help you cope in safe, healthy ways.

CCCs want to hear from YOU! Please tell the VHL Alliance about your experience at a CCC by completing this survey after each VHL-related appointment you have at your CCC: bit.ly/HowWasYourVHLvisit. VHLA will anonymously share your feedback helping CCCs maintain outstanding levels of care for VHL patients.

VHLA is looking for liaisons for each CCC. This volunteer opportunity requires ~5 hours/year. As a CCC liaison, you would be responsible for gathering feedback from VHL patients who attend the same CCC as you. Then, at least one time per year, you would meet with CCC Lead Team to share feedback and learn how VHLA can better serve the CCC. Check with Andrea to see if your CCC still needs a liaison by emailing her at: Andrea.Berkemeier@vhl.org.
The VHL Alliance has a challenge. We are tasked with making great progress in all areas of our mission. Over the past few years, VHLA has enhanced and expanded its efforts in education, support, clinical care, and research. Awareness is also at the core of our mission and is a major focus of 2017.

In 2017, we aim to increase awareness of VHL among the medical community and the general public. With more awareness of VHL, clinicians will improve diagnosis and treatment which impacts quality of life. Researchers will spur advances toward a cure. And the general public will better understand that finding a cure for VHL will lead to a cure for many forms of cancer.

VHLA continues to create partnerships with teams that have a wide reach and extensive visibility.

QuantiaMD is the largest social learning and collaboration platform for physicians with a reach of more than 225,000 physicians across the country. VHLA is working with QuantiaMD to create a series of short videos about the clinical aspects of VHL.

Elsevier’s Practice Update is an important educational resource for healthcare professionals connecting them with the information that matters most for patient care. VHLA is working to create a partnership that could serve as a “Journal Club” for healthcare professionals. Through the Journal Club, clinicians will have access to summaries of the most up-to-date information on VHL research and care.

VHLA is dedicated to research, education, and support to improve awareness, diagnosis, treatment, and quality of life for those affected by VHL.

Rare Disease Report (raredr.com/sap-partner/vhl-alliance) is a website and weekly e-newsletter with a reach of over 30,000 medical, scientific, investment, regulatory, and advocate professionals. VHLA has already posted several pieces of information on Rare Disease Report’s VHLA page. Please feel free to forward us your story or experience so it can be submitted to Rare Disease Report and help clinicians better understand what it is like to live with VHL.

Frontline Medical Communications reaches a wide range of medical specialties. This platform is being used to emphasize the importance of annual surveillance for managing VHL (mdedge.com/neurologyreviews/article/118235/rare-diseases/vhl-alliance-publishes-suggested-vhl-surveillance?channel=285).

The Mighty (themighty.com/category/cancer/von-hippel-lindau-syndrome/) maximizes personal stories to increase awareness of disease and disability. Stories are picked up daily by the world’s largest media outlets, driving a tremendous amount of awareness in the general public. While VHLA can suggest articles and information, stories submitted directly from patients draw interest and awareness. VHLA needs your help in getting out the word. We ask that you consider sending your narrative to The Mighty. We are happy to help you with the process including the writing!

It takes the entire community to increase VHL awareness. We can’t do it without you!

Patients and caregivers are invited to participate in VHLA’s “Better Together” peer mentoring program. This is a great way to share your experiences and learn from other people in the VHL community.

Please email Andrea.Berkemeier@vhl.org if you are interested in participating and minors (under age 18) will need parent/guardian consent to participate. Since the VHL community is spread out across the world, peer-mentor pairs will likely use long distance communications (email, phone, video) instead of meeting in person. Caregivers will be paired with caregivers, and patients will be paired with patients. Please note any special requests.
CONGRATULATIONS to William G. Kaelin, Jr., a leading VHL researcher for receiving one of the 2016 Lasker Awards. The Lasker Award is one of the most respected prizes in medicine. nyti.ms/2cA9fp5

UPCOMING

RUNdezvous couples run, February 11th, Newbury Park, CA facebook.com/RUNdezvous5k/

Weekend regional gathering in Minnesota in February us-mn@vhl.org

Michigan Chapter Meeting on April 22nd, Ann Arbor, MI us-mi@vhl.org

Monthly Florida coffee house meet and greets in the greater Tampa area us-fl@vhl.org

MAY AWARENESS EVENTS

Pancake breakfast in Michigan, May 6th us-mi@vhl.org

Adult-only comedy show/dinner fundraiser in Colorado, May 19th us-co@vhl.org

Team VHL 5k in Minnesota, May 20th us-mn@vhl.org

Concert event featuring The Farm Hands Gospel Quartet on May 25th office@vhl.org

RECENT

Warrior Run in Sligo, Ireland Over $12,000 raised!

5th Annual Cornhole Tournament in MA Over $10,500 raised!

7th Annual Cornhole Tournament in MA Almost $21,000 raised!

Happy Hour in NYC on January 12th $450 raised!

JUNE

9 - 11

YOUNG ADULT RETREAT
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.

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.

Transportation: Planning to fly? Use Nashville’s airport (BNA). Looking to carpool? Contact Andrea to see if other patients are coming from your area. Activities are planned from Friday night through Sunday afternoon in the greater Nashville area.

Fun and active events: Nashville outing, swimming, stargazing, hiking

Interactive lectures/discussion topics (tentative):

- New surgical/SRS/ablation techniques
- The logic behind screening guidelines: How were they established? How are they developing now?
- Life planning: transitioning to college/career, choosing a healthcare plan/disability, managing repeated surgeries and extended recoveries
- The adult patient: seeking a second opinion, participating in coordinated care, utilizing VHLA
- Family Planning: discussing VHL when dating, considering genetic inheritance and pregnancy for women with VHL

Check your calendar, choose your bathing suit, and email Andrea.Berkemeier@vhl.org to reserve your spot today!

CONGRATULATIONS to William G. Kaelin, Jr., a leading VHL researcher for receiving one of the 2016 Lasker Awards. The Lasker Award is one of the most respected prizes in medicine. nyti.ms/2cA9fp5
Emergency cards for VHL patients to carry in their wallets are now available.

As a VHL patient you are predisposed to having pheos which can cause life-threatening blood pressure spikes. Even if you have had your adrenal glands totally removed, we still encourage you to carry this card. If you end up in the ER and your doctor has never heard of VHL, this card will give them a quick overview of signs and symptoms associated with VHL manifestations.

Have an iPhone? We also encourage you to enter your specific information on the medical ID. To learn how to set up a medical ID on an iPhone, watch this short video: youtu.be/N_gqQHw8S_c.

Contact the office at office@vhl.org or call 800.767.4845 x4.

The VHL Alliance is looking towards the future. Would you like to join us?

By adding a few simple words in your will or trust, or by designating the VHL Alliance as the beneficiary of your retirement plan, life insurance policy, or commercial annuity, your gifts will save lives for generations to come. You can help VHLA dedicate itself to research, education, and support to improve awareness, diagnosis, treatment, and quality of life for those affected by VHL. Contact Heidi at Heidi.Leone@vhl.org for more information.
SAVE THE DATE!
2017 VHLA ANNUAL FAMILY WEEKEND

OCTOBER 13–15, 2017 — TAMPA, FLORIDA

Fundraising Dinner & Auction
Annual Family Meeting
Teen Event
Young Adult Happy Hour
2017 Team VHL 5K Run/Walk