VHL Alliance: Past, Present, and Future

As we reflect on the amazing year the VHL Alliance had in 2014, I want to personally thank you for your support. Because of your generosity, VHLA has been able to make significant strides with respect to research, education, and support for those affected by VHL.

In 2014, as part of our ongoing effort to identify a cure for VHL, VHLA funded $125,000 in grants for two unique research projects at Harvard Medical School and Tel Aviv University. In addition, we launched our own research program—the FDA-acknowledged, comprehensive Cancer in Our Genes International Patient Databank (CGIP, www.vhl.org/databank).

It is a “pivotal moment for the treatment of rare diseases”. “It [the databank] is being developed by NORD and the von Hippel-Lindau Alliance, which will help researchers searching for potential new treatments to learn more about the ‘natural history’ of the disease... We hope that this database will better serve the needs of patients with von Hippel-Lindau disease and help foster the development and testing of new treatments...”

— Margaret A. Hamburg, MD, Commissioner, Food and Drug Administration (FDA)

At the suggestion of our Research Council, CGIP was created. Now VHLA leads the way in clinical research for VHL, rare diseases, and other forms of cancer. This central database is maintained with the highest degree of confidentiality allowing researchers around the world to draw conclusions from hundreds to thousands of patients. While the database has only been in effect for a few months, researchers have already been able to glean relevant data. CGIP is leading the way in clinical research not just for VHL, but for rare diseases and cancer, too!

To help support our efforts, the VHLA raised nearly $590,000 in 2014. This record income allowed VHLA to support this innovative research. In addition, it has given us the confidence to aim for $674,000 for fiscal year 2015! With your generous support, so far we have raised over $250,000! We want to thank each of our donors for contributing to this amazing sum! While we still have a way to go, we are confident that we will reach on new goal.

While I am supported by Susan and Suzanne, a truly dedicated and hardworking team, we continue to rely on your support as well as the strong collaborations we have formed (e.g., NORD for the CGIP, Clinical Care Centers, Clinical/Research Advisory Councils, etc.).

Everyone in the VHLA community makes a difference! Please consider helping the VHL Alliance reach our goals in 2015! These goals include $674,000 in funding and reaching 1,000 CGIP database participants!

Susan, Suzanne, and I wish you a happy and healthy year and looking forward to serving you in 2015!

Annual Meeting Summary—Rockville, MD

The 2014 VHL Alliance Annual Conference was held in Rockville, MD on October 18th. The agenda included eight scientific presentations and a lecture on the Affordable Care Act, a talk on the State of VHLA, and two facilitated break-out sessions: one for VHL patients and one for family members and caregivers.

Dr. Pamela Stratton from NIH led off with a discussion of female health and pregnancy issues related to VHL. There have been no large-scale studies, so her conclusions are based upon the information currently available. Dr Stratton recommends that planning a pregnancy is the best way to ensure a healthy mother and baby. Studies suggest that VHL hemangioblastomas may be sensitive to the progesterin used in many birth control pills and devices, so a woman with VHL may want limit contraceptive choices to those that are non-hormonal or very low in progesterone. Planning when to become pregnant allows the woman’s health to be optimized by completing all imaging studies and addressing any significant findings prior to pregnancy.

Dr. Prashant Chittiboina from NIH discussed management of CNS hemangioblastomas. Hemangioblastomas are benign tumors which do not metastasize but can cause major medical problems for patients with VHL disease. In the Central Nervous System (CNS), they are seen in the back part of the brain, the cerebellum, the

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brainstem, and the spinal cord. The problems caused by hemangioblastomas are due to increased pressure which results from increased volume due to tumor growth, swelling, or bleeding. Surgery is recommended based upon symptoms, not the actual tumor growth. Removal will usually lead to a reduction in symptoms.

Dr. Wai Wong from NEI gave an overview of VHL in the eyes. VHL retinal lesions may cause no symptoms when the lesion is small and not leaking. However, if the lesion grows and begins to leak fluid, the surrounding retina can undergo swelling, distortion, and even detachment. When the retina is damaged, it loses its ability to detect light and vision is compromised. Rarely, in very severe cases, the entire eye can be affected to the extent that the intraocular pressure can increase (glaucoma) or become inflamed (uveitis) such that the eye may eventually need to be removed. The key to maintaining good vision is early detection and treatment of small retinal lesions. This is a reason to do early DNA testing in children and begin a retinal screening program before the onset of symptoms.

Dr. Karel Pacak from NIH presented information about pheochromocytomas. Although experts have not agreed whether the plasma (blood) or urine test is better, Dr. Pacak prefers plasma. When doing a plasma test, it is very important for the patient to lie down and rest 15-20 min after needle insertion before blood is drawn for the plasma test. Although there is lack of reference numbers for children, Dr. Pacak still prefers the plasma test. The urine test is more expensive and difficult to collect properly, especially when collecting samples from children. His take-home messages are: 1) Your doctor needs to understand the tests for pheos; 2) Any time you have surgery, it is important to let them know about pheos; 3) PET scan is becoming the way to diagnose head and neck paragangliomas.

Dr. Eric Jonasch from MD Anderson Cancer Center spoke about what is needed to advance VHL research to find a cure, we need: 1) New ways to either replace or repair defective VHL function; 2) Analysis of the other “broken” genes that conspire with VHL loss to cause tumors; 3) Model systems that replicate organ specific manifestations. The research grants awarded by VHLA for 2014 go to two researchers pursuing new approaches to cure VHL. Dr. Danny Segal is using an animal model (VHL-mutated zebrafish) to quickly test a number of currently available drugs to find if any are effective for VHL tumors.

Dr. Ramprasad Srinivasan from NIH spoke on the role of therapeutic drugs in the management of VHL. Surgery is the standard of care for tumors, but for VHL it is not curative. This means that systemic therapeutic agents should be used, when appropriate, to manage symptoms and delay surgery. Therapeutics must all have acceptable risks and side effects. A major obstacle to finding drugs to treat VHL is that recruiting patients is problematic. Most qualified VHL patients are leading “normal” lives and most drugs have disruptive regimens and side effects. An example is that over 200 patients qualified for a trial run by Drs. Srinivasan and Linehan at NIH, but the majority declined to participate. We need your participation in clinical trials in order to find effective therapeutic treatments for VHL.

Dr. Adam Metwalli from NCI discussed management of VHL-associated kidney cancer. He explained how removal of kidney tumors prior to the metastatic stage “resets the clock.” The average time between surgeries is 50 months following a partial nephrectomy and the need for dialysis is avoided in most cases. Using partial nephrectomy, Dr. Metwalli and his colleagues have found minimal change in pre and post-op renal function. He also addressed the option of radiofrequency ablation (RFA) instead of open surgery. Surgery is actually more nephron-sparing, more accurate, and allows removal of more tumors. Another disadvantage of RFA is that this method does not “reset the clock” as much as a through partial nephrectomy, meaning that there will be a need for another procedure to remove kidney tumors sooner. RFA is also less likely to spare kidney function due to fibrosis following the procedure.

Dr. Peter Choyke of NCI spoke about imaging used to manage VHL. Magnetic resonance imaging (MRI) uses magnetic fields, not ionizing radiation. This means that MRIs do not add to your lifetime radiation exposure. There are two primary drawbacks of MRIs: patient claustrophobia in the closed units, and incompatible implanted devices (such as some heart pacemakers). Computed tomography (CT) scans were used in the past for abdominal imaging. The problem is radiation exposure. Use of contrast agents has resulted in reduced radiation while maintaining image quality. Ultrasound is another option, but the results are dependent upon the technician performing the test, so for most VHL tests, MRIs and CT scans are preferred.

Diane Dorman from NORD spoke about the ways in which NORD works with the entire rare disease community to advocate for changes at the federal government level. She emphasized that the Affordable Care Act (ACA) which prevents denial of health insurance coverage based upon pre-existing conditions and eliminates annual or lifetime financial caps on coverage, is only the latest legislation which has benefitted those with rare diseases. Although the ACA is a federal regulation, the actual insurance available varies by state, so it is important to visit: https://www.rarediseases.org/ NORD worked with the FDA to develop the Orphan Drug Act (lower regulatory requirements for drugs developed to treat rare diseases), and The Genetic Information Non-Discrimination Act (health insurers and employers cannot discriminate on the basis of DNA test results). NORD is now developing a Rare Action Network in order to work with patients to advocate at a state level.

A more in-depth summary of each presentation, audiotapes, and slides from the presentations may be viewed on the VHLA website:

http://tinyurl.com/2014-rockville-md
Join the VHLA Coin Drive!
How much can you collect by June 2015?

Look in your pockets, the creases of your sofa, in the car, and even in the washing machine. Collect them in a jar. Even ask your friends and neighbors to collect their change. You’ll find that it really adds up.

Trade it in for dollars or a check and send in what you have collected by June 15, 2015 to the VHL Alliance, 2001 Beacon Street, Suite 208, Boston, MA 02135.

Clinical Care Center Updates

The new VHL Clinical Care Center standards were announced to our existing Clinical Care Centers this fall, and there has been a very positive response. (For more information visit www.vhl.org/ccc.) A number of current Clinical Care Centers applied to be recognized as Comprehensive Clinical Care Centers. In addition, there was interest from new hospitals recommended to us by VHLA constituents. Your physician and hospital recommendations are key to expanding this valuable resource. Please contact wellness@vhl.org with any suggestions or feedback about any of the centers.

New Comprehensive Clinical Care Centers
• Cleveland Clinic—Cleveland, OH, adult + pediatric
• Columbia Presbyterian—New York City, NY, adult + pediatric
• MD Anderson—Houston, TX, adult + pediatric
• Stanford—Palo Alto, CA, adult only
• University of Chicago—Chicago, IL, adult + pediatric
• University of Pennsylvania—Philadelphia, PA, adult + pediatric
• University of North Carolina—Chapel Hill, NC, adult + pediatric
• University of Virginia—Charlottesville, VA, adult + pediatric
• Yale University Hospital—New Haven, CT, adult + pediatric

NEW Clinical Care Centers
• University of Minnesota—Minneapolis, MN, adult + pediatric
• Hospital Italiano—Buenos Aires, Argentina, adult + pediatric
• Hospital Privado—Cordoba, Argentina, adult + pediatric
• Peking First University Hospital—Beijing, China, adult + pediatric

If you have been diagnosed with VHL and have relatives in China, please contact the hospital. They will work with patients throughout the country to test for VHL.

International VHL Symposium for Young Adults, 2015

July 31–August 3, 2015, Utrecht, The Netherlands

VHL Europa is very pleased to announce the International VHL Symposium for Young Adults 2015. This is the first time this event is being organized, by host country, The Netherlands.

This is an opportunity to network and share global experiences with VHL. Important choices need to be made as a young adult and, from a psychological point of view, sharing experiences is extremely valuable.

The program will offer a balance between leisure activities and serious topics, including: VHL Quiz, interactive VHL-related lectures, BBQ, Festival ‘De Parade’ Utrecht, and Jordaan tour in Amsterdam.

Criteria to participate: Must be 18-27 years and VHL-affected. Should have sufficient English knowledge to be able to express themselves. No parents and no partners policy.

Any questions? Just4youth@vonhippellindau.nl

We hope to meet you in July 2015!
—Barbara Bezemer, The Netherlands

VHL Programs Available for You

• Monthly conference calls for patients and caregivers: http://tinyurl.com/telephone-discussions
• VHL Partners: Mentoring and support groups for patients and caregivers. http://tinyurl.com/vhl-partners
• Annual Screening Reminder Tool: You sign up and it will automatically remind you when to get your next screening test. http://www.vhl.org/wordpress/vhl-screening-reminder
• New Clinical Care Centers (CCC) and Comprehensive Clinical Care Centers (CCCC). VHLA has added more Clinical Care Centers and have categorized several new and existing CCC’s into Comprehensive Clinical Care Centers. http://vhl.org/ccc

The VHL Alliance is your community of caring and support that is always available to you during the roller coaster moments in your life. You can always call the VHLA office at 1-800-767-4845 ext. 4 or write to office@vhl.org.
VHL Patient Vignettes — Personal stories, thoughts, and tips submitted by people dealing with the diagnosis of von Hippel-Lindau

Life with VHL has many ups and downs. This newly-published book was compiled from interviews of patients and caregivers dealing with the day-to-day challenges of living with VHL or a loved one with VHL. It is great for those new to VHL and inspiring to everyone. Below are some excerpts.

“...While I worried that I was going to develop another brain tumor, I realized that, except for some minor annoying side effects, I was living my life normally and without restrictions.”

“...When I found out I had VHL, I started crying. I felt depression, anger, and fear.”

“...The more knowledge I have about tumor size, rate of growth, and symptoms to look for, the more it helps me feel in control of the situation.”

“...You have VHL. VHL does not have you.”

VHL Patient Vignettes is available for purchase for $6.50 from the VHLA website: http://www.vhl.org/patients-caregivers/resources/vhl-patient-vignettes/

11th International VHL Symposium, Madrid

The 11th International VHL symposium in Madrid was an exciting, informative meeting with presentations by researchers and clinicians on the latest findings in VHL therapies and basic research on the biological mechanisms behind VHL tumor formation. The material was presented for an audience primarily of academic and clinical researchers. Selected slide presentations are posted on our website with a short summary of the talks, including the talk below, at: http://tinyurl.com/madrid-symposium.

Screening and diagnostic aspects of Endolymphatic Sac Tumors (ELSTs) – Marie Louise Mølgaard Binderup; University of Copenhagen, Denmark

ELSTs occur in up to 16% of VHL patients and 11–30% of those are bilateral. Most patients (96%) are not diagnosed until they have symptoms: hearing loss (91%), ringing in the ears (64%), or dizziness (52%). These symptoms are not always cured by surgery, so the challenge is to diagnose at the earliest possible stage.

Hearing loss from ELSTs can be gradual or sudden:
Our Sincere Thank You for Donations From

Donations are seen on the printed version only.

Your help is greatly needed

We also need volunteers. Call 1-800-767-4845 ext. 4
MARK YOUR CALENDARS NOW!

2015 FUNDRAISING DINNER & AUCTION
Friday Evening, October 16, 2015
Chicago, Illinois

HONORING
Sarah Nielson, MS, CGC, University of Chicago, IL
Janice and Scott Capinegro, Barrington, IL

2015 VHL ANNUAL FAMILY MEETING
Saturday, October 17, 2015
Chicago, Illinois

2015 TEAM VHL 5K RUN/WALK
Sunday Morning, October 18, 2015

Details will be announced as they become available