Restructuring VHLA’s Clinical Advisory Council

by Eric Jonasch, MD, VHLA Board of Directors; Chair, Clinical Advisory Committee and Research Council

As chair of VHLA’s Clinical Advisory Council, I am very excited to inform VHLA’s constituency of the recent restructuring of this important group of clinicians. With an eye on improving care for patients with VHL throughout the country and around the world, we have recruited leaders in Urology, Endocrinology, Ophthalmology, Clinical Cancer Genetics, Medical Oncology and Psychiatry/Psychology to the Council. See list on page 7.

The major tasks of the Clinical Advisory Council are to assess and update best practices in each of the organ specific domains, vet and rate aspiring and currently established Clinical Care Centers, provide a resource for education of the broader community, enhance visibility of VHL disease in various academic forums, review and update the current educational resources available for physicians and patients, and identify clinical research questions that will benefit from collaboration both within and across disciplines.

Each of the disease groups is currently working on reviewing and updating the literature in their respective fields. Where appropriate, groups are generating review papers which will be published in organ and disease specific journals, with the goal of reaching as large an audience as possible within each subspecialty. Once all of these reviews are complete, an overall review article will be written which will reference the organ-specific literature and will form an entry point into what will then be fully current information on best practices for the management of VHL. Our goal is to make it as easy as possible for health care professionals who encounter patients with VHL to take care of them, and to encourage referrals to centers of excellence where appropriate.

The Clinical Advisory Council is also working hard to strengthen the network of Clinical Care Centers that can provide reliable, cutting edge diagnostic, screening, and therapeutic options for individuals with VHL. Clinical Care Centers are reviewed every two years to make sure that the necessary health care providers and resources are available to deliver the best possible care. A list of Clinical Care Centers is available in the VHLA website at www.vhl.org/ccc.

The long term goal of the Clinical Advisory Council is to improve care for patients living with VHL. To that end, various members of the Council are working with partners in the pharmaceutical industry and with VHLA’s Research Council to identify new ways to screen and treat the various manifestations of VHL. Our goal in the next ten years is to dramatically reduce the number of operations and procedures patients have to undergo. We are optimistic that this is an achievable goal and will work hard to make it a reality for the VHL community.

Welcome Heidi A. Leone, New VHLA Director of Development

Heidi joined the VHL Alliance team during the summer of 2015, as Director of Development. Her lifelong passion to fight against cancer and her diverse fundraising skills make her uniquely suited for the mission of VHLA. She has worked in non-profits for 20 years, specializing most recently in annual giving and major gifts. Her background with national non-profit organizations, such as the American Cancer Society, and her varied responsibilities with independent schools, faith-based organizations, and children, have prepared her to lead in a variety of settings and fundraising venues.

Heidi brings along with her enthusiasm, passion, and generosity of self in her career as well as her private life. She spends as much time as possible with family, friends, and her Jack Russell Terrier.

Her other interests include cooking, tennis, gardening, reading, and being by the water.
Living with VHL from the Young Adult Perspective

by Lindsay Schmidt, MS, CGG

Von Hippel-Lindau syndrome affects patients in various ways. While most research has focused on the genetics of VHL and medical implications of the syndrome, social and emotional experiences of VHL patients have received far less attention. Additionally, there is a lack of literature addressing the experiences of young adults living with VHL. This lack of knowledge served as inspiration for my thesis study, which I completed at Brandeis University while earning my master’s degree in Genetic Counseling.

The purpose of this study was to explore the social and emotional aspects of living with VHL as a young adult. My investigation was performed by conducting telephone interviews with ten young adults (18–26 years old) living with VHL in the US. After interviewing all participants, I identified five overarching themes to which many VHL patients will relate.

Living with Uncertainty

Many participants expressed that living with uncertainty is a challenging aspect of living with VHL. As Sophie pointed out, a VHL diagnosis leads to many ‘what-if questions’, and participants seemed to struggle with if, when, and how VHL will affect them. Tumor growth, timing of VHL manifestations, and VHL’s impact were concerns.

“Every time I go to NIH, I just never know what I’m going to find out; is there anything new or is anything getting worse?” — Peter

Maintaining a Positive Attitude

Despite living with continual uncertainty, many participants seemed to maintain a positive attitude. Several participants positively framed their VHL experiences while others exhibited perseverance.

“Experiences that you go through either make you better or they make you bitter. I wanted them to make me better, and I think that they have. ... it has ... been a really awesome growing experience for me that I’m lucky enough to have.” — Julie

Significant Means of Support

Participants reported large support systems, which often included a combination of family, romantic partners, friends, coworkers, and health care professionals. However, when it came to VHL-related concerns, single participants most often reported their mothers as their strongest confidants, while married participants most often reported that they would turn to their spouses first.

Polarizing Effect on Relationships

We found that VHL can have a polarizing effect on relationships, increasing the closeness of some and the distance of others. This effect seemed to apply to different types of relationships, including familial, romantic, and friendships.

Overall, participants provided more examples of how VHL has brought them closer with others than how VHL created distance within relationships.

“I didn’t share as much personal stuff with [my mom] before, and now she is one of my best friends. I tell her everything.” — Seth

Impact on Life Decisions

Young adults often face major life decisions, and we found that VHL can have an impact on decisions regarding location, careers, and childbearing. Living with VHL impacted where two participants lived, influenced three participants to pursue careers in the health care field, and led to childbearing concerns for nine participants.

“I see the fun and the joy [a child] could bring. But a 50 percent chance of passing this VHL onto any kids I have is way too high of a percentage for me to really consider that.” — Scott

Assisted reproductive technology was discussed with the participants as a way to prevent having a child with VHL and half were open to utilizing in vitro fertilization (IVF) with preimplantation genetic diagnosis (PGD), while two participants did not support the use of this technology. Two of the five participants who were open to IVF and PGD were not aware that these technologies existed until mentioned during the interview; neither of them had received genetic counseling in the past.

In conclusion, there is more to VHL than tumor development and surgeries. It leads to perpetual uncertainty, can affect relationships (in both positive and negative ways), and is often a consideration in life decisions. Those who fall within this young adult age group would likely benefit from establishing a long-term relationship with a genetic counselor. They can be the guide to finding ways to manage uncertainty, recommend VHL support groups, and provide information about available reproductive options.

The hope is that this study will spark the interest of researchers and provide inspiration for larger and more focused studies of this neglected age group, with the ultimate goal of providing more comprehensive care for VHL patients.

Note: Participants’ names have been changed to protect their privacy.

Lindsay is a recent graduate of Brandeis University’s Genetic Counseling Program. Her research will be presented at the National Society of Genetic Counselors conference in October, 2015.

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Go to smile.amazon.com and choose VHL Alliance as your charity. Go there every time you shop at Amazon!
How I Became Proactive with VHL
by Ellen W, VHL patient

When I was diagnosed, I felt like so much of it was out of my control. Though I could not control the results of the tests, there were a few things that I could do to make living with VHL a little bit easier.


Next, I decided to get involved. The VHL Alliance has a plethora of support systems set up for VHL patients and families/friends. Since there was no chapter in Illinois, I decided to become a regional chapter leader to connect with others in my area. I also joined the Facebook group and Inspire. I realized I had a support system who understood the emotional roller coaster and challenges of being faced with a VHL diagnosis. Much of this information I found here: www.vhl.org/patients-caregivers/resources.

Increased Incidence of Oral Ulcers/Canker Sores within the VHL Community

By Emily Walsh, Erika del Castillo, and Jacques Izard, The Forsyth Institute, an affiliate of Harvard School of Dental Medicine.

Jacques Izard, PhD is a member of VHLA’s Research Council.

A recent finding from the Cancer in our Genes International Patient (CGIP) Databank indicates a higher rate of oral ulcers or canker sores (clinically known as aphthae, stomatitis, aphthosis) in the mouths of VHL patients. Canker sores are small, often painful inflammatory ulcers that are located inside the mouth or on the gums. They range in size and severity – from the presence of small singular oval ulcers (<1 cm in size) to larger ulcers, which can persist for months at a time.

The underlying cause(s) of canker sore formation within the VHL community is still not known. Current treatment primarily revolves around pain management and anti-inflammatory medication.

I also needed to focus on financials. The numerous medical bills and Explanation of Benefits (EOB) were overwhelming. I made a binder to organize my bills and EOBs by date of service for referencing and tracking. I investigated taking advantage of a Health Savings Account (HSA): http://tinyurl.com/treasury-gov-healthsavings. Another option I was made aware of was to check out hospitals and doctors’ offices for financial assistance programs.

Looking at all the opportunities for research, I am participating in the Cancer in our Genes International Patient (CGIP) Databank. There are a multitude of surveys to help researchers understand the “natural history” of VHL. It also provides a unique opportunity to understand what factors impact tumor development and growth rate. www.vhl.org/databank.

I continue to learn. As science and research evolves, so do the treatments and information surrounding VHL. I truly discovered that knowledge is power! www.vhl.org/patients-caregivers/basic-facts-about-vhl.

I have hosted and participated in meetings and fundraisers. It is important for me to raise awareness, and it is vital for funds to be raised to support research. I found some fundraising ideas here: www.vhl.org/ways-to-help/fundraising-ideas. This year I will be attending the Annual Meeting, the Fundraising Dinner & Auction, and the Team VHL 5K Run/Walk in Chicago. I hope you will join me! www.vhl.org/meeting.

Image: Wikimedia Commons

So why is VHL associated with canker sores?

YOU CAN HELP US FIND THE ANSWER!

www.vhl.org/databank
Drug Research and Development

The drug development process in the US can be a lengthy process as multiple steps are required in order to ensure that approved drugs work for their intended purpose with a minimum of side effects. When a drug is developed for a relatively common disorder such as high blood pressure, the pharmaceutical company must follow the standard FDA protocol.

In the case of a rare disorder, such as VHL, it is usually not possible to find enough patients who meet the study criteria and are willing to be subjects. This fact, along with the fact that there is only a small market for the drug, means that developing drugs specifically for rare diseases may not be financially beneficial to the pharmaceutical companies. This barrier to drug approval for rare diseases was removed in 1983 with the passage of the Orphan Drug Act. Applications for FDA approval of new drugs under the Orphan Drug Act have grown quickly.

While the Orphan Drug Act decreases the number of patients required for a clinical trial and shortens the time for FDA review and approval of a potential medical therapy, it does not impact the time it takes to discover the treatment (basic scientific research) and move it from the lab bench to patient. This still takes many years!

Orphan drugs are defined as those developed for disorders with less than 200,000 patients in the US. Any drug developed for VHL will be in this category as there are only about 100,000 people in the US diagnosed with VHL.

The VHL Alliance is actively involved in several important steps needed to find drugs that will manage the many aspects of VHL. The first step is to document the natural history of VHL. This knowledge is needed in order to determine if any drug being tested is making a difference in the course of the disease. The CGIP Databank is the way that we will learn the natural history of VHL. It is important for everyone to participate and document their individual experiences. The information is key to shortening the drug approval time.

Research grants from VHLA go to support basic research in the mechanisms of VHL tumor formation. A thorough understanding of the tumor formation process is necessary to develop drugs to treat VHL. The VHL gene is also part of tumor formation in patients without VHL, so this knowledge will potentially benefit everyone with cancer. The potential application of drugs developed for VHL to a wider group of cancer patients is another incentive for pharmaceutical companies.

Hatfield-McCoy Marathon

On Saturday, June 13th, Team VHL participated in the the Hatfield McCoy Marathon and, at publication time, raised $20,375 for VHLA and more is expected to come in. For pictures and to add your donation, please see: http://tinyurl.com/hatfield-mccoy-marathon-vhl

2015 Rick Bracey 29er Tournament

The entire 29er Tournament Committee would like to thank everyone for their contribution to the Rick Bracey 29er VHL Fundraising Softball Tournament held on June 13th, 2015, in Surrey, BC, Canada, which raised more than $24,050 in a single day! Cumulatively, this tournament has now raised more than $124,000 for VHL research and education!

Mom’s Day Run 2015

Thank you to everyone who participated in the Mom’s Day Run! It was huge success, and we couldn’t have done it without you. Emily B sang the National Anthem! $4,836 was raised for VHLA.

Van Cortland Track Club Race for VHL

June 18th, Bronx, NY: Andrea R organized a race to benefit the VHL Alliance and raised $1,050.

Charity Magic Tournament-LEGACY

Josh C sponsored a Magic Card Tournament on April 25th in Bellevue, WA, and raised $1,476

Happenings at VHLA

Bless This Mess

May 14th: VHLA Board Members Manuel Greco and Bettina Micheli hosted “Bless this Mess—do the #vhllwarrior dance” in Brooklyn, NY. $4,410 was raised.

MAY AWARENESS EVENTS

VHLA Coin Drive: Thank you to all who participated! It’s never too late to join the cause. http://tinyurl.com/vhlcoindrive

Cocktails for a Cause, Phoenix, AZ, May 9: Karen R & Bobby K raised $6,018.

Bake Sale, Ava, MO, May 9, 2015: Vanessa L raised $161.

E-Waste and Shred Day Event: Miller Recycling, May 16, Plainville, MA: Thanks Maria R! $425 was raised.

Northern California Regional Meeting: The Northern California Chapter hosted a regional meeting at Mission Pizza in Fremont, CA, on May 31st. About 20 people had pizza and drinks while participating in discussions relevant to VHL. Thank you Greg T and Jackie G!

Thank you to Anna H and alter ego Beatrice Steinberg for their amazing Social Media VHL Awareness Campaign and to Emily B for her awareness video! We appreciate everyone who participated in VHL May Awareness Month!
Support VHLA Through the United Way
Check with your local United Way at unitedway.org for details on how to support VHLA in this way!

2015 CFC Campaign
Are you or someone you know a State or Federal government worker?

The 2015 CFC Associated State Campaigns are underway. Your donation to the VHLA can be deducted directly from your paycheck. You can fund VHLA under its DBA: Cancer Research and Assistance - VHL. Federal ID# 10934.

Welcome New Board Members

Barbara Correll has more than 15 years of experience in health care marketing and communications, including work in brand building initiatives, corporate positioning, public health campaigns, public relations, and product launches. She is currently the Senior Vice President of Marketing at StayWell, a provider of patient education and population health management solutions.

Earlier in her career, she worked at the AMA, where she managed public health campaigns and programs. Barbara earned her BS in Exercise Science & Nutrition and an MS in Health Care Administration and Policy. Barbara learned about VHL and VHLA through a pro bono marketing engagement. She is excited to continue to contribute her marketing and communications experience to help raise awareness about VHL.

Heidi Larson comes from a medical background. In 2008, she learned that she has VHL. It is the continued work of the VHL Alliance that has set up a protocol and monitoring schedule for patients with VHL and their families and, because of this kind of surveillance, she believes her best years are in front of her.

After receiving her BS in Nutrition and MA in Teaching, Heidi became a lifestyle, nutrition, and fitness instructor for over 20 years. She has had extensive medical clinical and medical sales consulting plus 15 years’ experience in clinical medical practice management and patient care.

Anoop Mangat is a senior executive and software architect with over 15 years of experience in global enterprise systems for the financial services industry. He is an intuitive problem solver with excellent management, technical and presentation skills.

Anoop is currently an Executive Director at Morgan Stanley and is cofounder/CTO of Searchspace (now Fortent/Actimize). Prior to moving to New York, he was a Research Fellow in the Intelligent Systems Laboratory at University College, London.

Anoop is a good friend of Manuel Greco, Vice Chair of the VHLA Board of Directors, and is personally invested in furthering the cause of the VHL Alliance.

Patricia Tang is an attorney and real estate broker in New York City. Her commitment to philanthropy is evidenced by her active support of the VHLA and other causes. Patricia also sits on the board of Best Buddies New York and Savvy Ladies, both organizations helping those in need.

Patricia’s mother and brother were diagnosed with VHL. She is immensely grateful to the VHLA for the counseling and support she received as she confronted her own genetic predisposition to the disease and attempted to navigate the difficult process of genetic testing.

Patricia is committed to raising awareness about the disease, finding a cure, as well as supporting others who seek practical information that will assist sufferers of VHL and their families.

Anna Waller has worked in sales for 30 years and is currently selling a mobile app for Black Book. She received her MBA in 2006.

Anna is an avid reader and creative writer. She lives in Georgia with her husband, John, of 32 years. They have 3 children and 2 grandchildren. Her youngest daughter was diagnosed with VHL in 2008 at the age of 17 and is the first in the family with VHL.

By joining the VHLA Board, Anna hopes to contribute to its growth and expansion. She has attended the last two VHL Alliance conferences and enjoys the monthly telephone discussion groups.

To see all of the VHLA Board Members and to see more detailed bios, please go to http://tinyurl.com/vhla-board

Fundraising While Shopping Online
iGive July Contest Means Bigger Donations for VHLA

Tell your friends. Each person who joins iGive in July for the first time can translate to $5 automatically donated to his or her cause ... no purchase necessary! All they need to do is try the iGive Button through 10/15/15.

The person who gets the most new members to join iGive in July and try the button through 10/15/15 earns $500 for his or her cause. $250 prizes go to the second and third place winners’ causes.


Help VHL Alliance every time you shop at 1,587 of the most popular online stores: http://tinyurl.com/igiveVHL.

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Chicagoland, Illinois

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

www.vhl.org/meeting/dinner2015

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

www.vhl.org/meeting/meeting2015

2015 TEAM VHL 5K RUN/WALK
Sunday Morning, October 18, 2015
Hyatt Regency Schaumburg, Illinois

www.crowdrise.com/teamvhl2015