

VHLA: Plans for 2014

At the VHL Alliance, we are always working to find ways to better serve our constituency. Challenges being faced by VHL patients and their caregivers are often identified through VHLA's Facebook (www.facebook.com/groups/VHLawareness) and Inspire discussion groups (www.inspire.com/groups/vhl-alliance).



One concern frequently expressed through these venues is the emotional rollercoaster of living with VHL. Ups and downs with anyone, directly or indirectly, impacted by a long-term medical condition such as VHL, are inevitable. Anxiety may be triggered by events such as the diagnosis, preparing for annual scans, decisions on when to operate, and thoughts about future health.

Complicating the emotional rollercoaster is the feeling of isolation and loneliness. Is there anyone who can actually understand? How do others with VHL cope? What can I learn from them?

VHLA's online support communities help meet some of the needs of many people impacted by VHL. These are international discussions on living with VHL. One can learn from the experience of others while providing and receiving support and sharing tips for dealing with the emotional rollercoaster.

The question is: "What more can VHLA do to help people during those challenging times." Are there more programmatic efforts or website content that VHLA can offer?

As we begin 2014, we have decided that finding answers to dealing with the emotional rollercoaster will be a major piece of VHL Alliance's work for the upcoming year. What services need to be created? What training for staff and Chapter Leaders and Hotline volunteers should be implemented? Are there self-help tools that can help our constituency deal with the emotional rollercoaster of VHL? Should discussions about the emotional rollercoaster be included in a patient's annual screening? And how can we help family members with their feelings of anxiety?

As always, the best way for us to learn is by hearing from you, our constituency. Please call (800-737-8845 x4) or email us (director@vhl.org) with your ideas and thoughts. We are always here to listen!

Worry does not empty tomorrow of its sorrow,
it empties today of its strengths."

— Anonymous

Appealing Insurance Denial for Screening Tests

The following article was written for you to share with any of your physicians who have ordered screening tests to help manage your VHL disease and monitor any lesions. The technical wording and "insurance-ese" will assist you and any of your doctors who run into a denial for a screening test and need to appeal this decision. Please notify wellness@vhl.org if you are having issues with insurance coverage for screening tests as we want to document this.

An all too common challenge faced by VHL patients is a denial by their health insurance company for a recommended screening test. The reason for this denial is given as "lack of medical necessity." An informed VHL patient working with their doctor can educate the insurance company and demonstrate that it is in the best interest of both the patient and the insurance company to cover recommended screening tests. Screening will find any problems in the early stage which is the most successful and least expensive time for treatments. Improved surveillance guidelines have increased the life expectancy of individuals with VHL by over 16 years since 1990 [Wilding *et al*, 2012].

Insurance companies, HMOs, Medicaid/Medi-Cal, and Medicare have guidelines in place for determining "medical necessity." Each insurer may develop their own guidelines, follow the policies of their umbrella association (ex: Blue Cross Blue Shield), or contract with a company that creates "utilization management" guidelines. The claims reviewers deciding upon medical necessity for specific claims are often advance practice nurses working with a physician medical director. They are not experts in rare genetic diseases and may deny something that they do not understand. They can be persuaded, though, by well-accepted national guidelines and reviews such as: USPTF (US Preventative Services Task Force) recommendations, NCCN (National Comprehensive Cancer Network) recommendations,

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Appealing Insurance Denials for Screening Tests

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or even *GeneReviews*. For example, here are the VHL recommendations from *GeneReviews*: <http://www.ncbi.nlm.nih.gov/books/NBK1463>.

Another important topic is evaluation of blood relatives of diagnosed VHL patients. The American Society of Clinical Oncology (ASCO) identified VHL as a Group 1 disorder (a hereditary disease for which genetic testing is considered part of the standard management for at-risk family members) (American Society of Clinical Oncology 2003). It should be noted that current DNA tests do not detect VHL mutations in all at-risk individuals. This means that diagnostic genetic testing of the VHL gene should be considered a standard of care for each person in whom VHL is suspected. If a VHL genetic mutation is identified, then each of their first-degree relatives (parents, siblings, and children) should be tested for that specific mutation. Ideally, a genetics professional—a genetic counselor or a clinical geneticist—should be involved in the genetic testing process by providing pre-test evaluation and counseling, test coordination, and post-test counseling.

Insurance claims reviewers may not have any idea where to look for documentation to support a claim outside of their standard guidelines. This means that the burden will fall to your physician ordering the screening or diagnostic test(s) to do several things. First, before a patient is sent off to have a test or a procedure, contact the insurance company and get pre-authorization. If the insurance company balks, then the physician should write a letter of medical necessity in which a persuasive argument is made that the requested procedure has important clinical utility—*i.e.*, the results will cause a real change in patient care. Clinical practice guidelines, vetted by a national organization—like NCI (National Cancer Institute) or the AAP (American Academy of Pediatrics)—carry weight and should be cited in the letter of necessity as a way of convincing the reviewer that the request should be approved because to not do so would mean failure to follow the clinical standards, thus not allowing the physician to meet the legally required standard of care.

If the reviewer still refuses to approve the test or procedure, and your arguments are strong, you can request, in writing, that the insurance company have the request reviewed by an outside reviewer. You can also ask that that reviewer have certain qualifications, such as board certification in a particular specialty, such as Clinical Genetics or Neurosurgery. The insurance company or government payor must comply with these requests.

Naturally, this is tiresome, inefficient, and expensive for you, your doctor, and your health insurer. However, this gives you the best chance to remove the barrier to having your claim approved and will allow you to manage your VHL disease according to the latest guidelines. Your efforts will also ease the way for other VHL patients having the same health plan to obtain approval for needed screening tests.

Please let the VHL Alliance know if you run into difficulties with insurance denials.

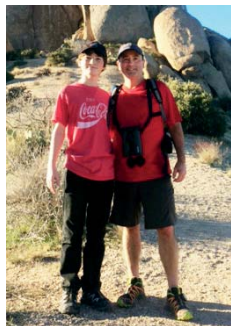
10 Things No One Can Steal From You

Excerpts from <http://www.marcondangel.com/2012/09/21/10-things-no-one-can-steal-from-you>

Of all the things that can be stolen from you—your possessions, your youth, your health, your words, your rights—what no one can ever take from you is...

- 1. Your uniqueness.** You are unrepeatable. There is a magic about you that is all your own. You are always amazing just as you are.
- 2. How you feel about yourself.** It takes a long time to learn how to NOT judge yourself through someone else's eyes, but once you do, the world is yours for the taking.
- 3. Your intuition.** Follow your intuition. Never compare your journey to someone else's. What you're looking for is already somewhere inside you.
- 4. Your passion.** The moment is now to find your passion, whatever it is. Even if things don't work out as planned, you'll still be heading in the right direction.
- 5. Your determination.** When life gives you something that makes you feel afraid, that's when life gives you a chance to grow strong and be brave. Nothing is permanent, not even your mistakes, failures, or troubles.
- 6. Your attitude.** Something nobody can take away from you is the way you choose to respond to what others say and do to you. Choose to be happy and positive.
- 7. Your ability to spread love and kindness.** The measure of your life will not be in what you accumulate, but in what you give away.
- 8. Your hope.** It's not the load that breaks you down, it's the way you carry it. Hope is the little voice inside your head that whispers "maybe" when it seems like the whole world is shouting "impossible!"
- 9. Your knowledge and life experiences.** Character and wisdom are sculpted over time. They come with loss, lessons, and triumphs. The seeds of your success are planted in your past failures.
- 10. Your choice to move on emotionally.** Don't let past relationships and old mistakes ruin your future. Learn the lesson, release the pain, and move on.

Calling All Teens!



by Gunnar K, Arizona

Not every thirteen year old can say they've had brain surgery! Mine occurred this past July. Hearing the news that I had a brain tumor was very surprising, but I had seen my dad go through VHL surgeries, and that really helped. My hope is that by sharing my experiences with other teens, I can help others deal with similar situations.

I just recently had a second surgery, this one for retinal blood vessel tumors in each eye. The good news is that I'm fine. In fact you wouldn't even know what I've been through just by looking at me. Thankfully I've recovered fully without any lasting issues. Having to deal with lots of doctor appointments and routine tests like MRIs and ultrasounds is just part of my life with VHL.

I was diagnosed at the age of 8 through genetic testing. Knowing that I had VHL probably saved my life! We did the MRI on my brain and spinal column when I reached the teen years, following the VHL Alliance protocols, and that's what identified the blood vessel tumor in my brainstem. Thank goodness for having proactive parents and a great neurosurgeon!

Today I'm just another busy teenager...playing tennis and video games, running and hiking with my parents around Phoenix, chatting on Instagram and keeping up with school work. Does homework ever end? I know that any teenager reading this has their own life issues and experiences. They may be similar or different than mine, but the fact remains that we all go through a lot during this phase of our lives. Sometimes it helps us just by sharing our experiences or helps others to hear they are not the only one going through a life situation. I'd like to help kick off a teen forum for those living with VHL. Some of you have it and some live with others who do. Would you be interested in joining me? The contest below can get you started!

Teen Writing Contest

TOPIC: "Life is hard even when you don't have VHL. How do you deal with teenage life?"

Send your article to teens@vhl.org

The winner receives a \$50 iTunes gift certificate

ASK THE EXPERTS:

Tumor Growth Rate and VHL

Ramaprasad Srinivasan, MD, PhD
Head, Molecular Cancer Therapeutics
Urologic Oncology Branch
National Cancer Institute

Dr. Srinivasan is a medical oncologist working with urologists at the NCI. He is active in creating the Clinical Trials Task Force. His goal is to explore ways to medically manage VHL with the objective of replacing or delaying surgery with drug treatments.

The best practice in caring for VHL patients is to minimize the number of surgeries while preventing metastatic disease in order to allow the organs, such as the kidneys, to continue to function. Tumor growth rates vary, even in the same person. An individual can have different rates at different times as well as different rates between tumors in different organs. This makes it very difficult to predict tumor growth, especially 5 to 10 years into the future. Clinicians use their best estimate based on their clinical experience.

The measurements referred to in the tumor growth studies are based on the "long diameter" of solid tumors. It is likely that eventually software will be developed that allows us to measure tumor volume, but due to their irregular shape, all studies and guidelines currently use the longest diameter measurement.

A tumor "long diameter" (the longest dimension)

measure of 3 cm or greater generally needs surgical removal. A faster growth rate may indicate a need for surgery to remove a smaller tumor. If the tumor is predominantly cystic, then surgery may be able to be delayed beyond the 3 cm stage.

Other factors to consider are: number of surgeries, kidney function, presence or absence of metastatic disease, and previous tumor growth rate. Basically, ask "What is gained by doing surgery now?" It is still unknown what makes tumors return or metastasize at the molecular level. However, larger tumors are more likely to have factors that allow them to spread.

The first paper on tumor growth rate from NIH came out in 1992 (<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1303573/#bib46>) and found that growth rates ranged from 3 mm to 1.6 cm per year, and that the average (mean) growth rate is 3–4 mm per year. (1 mm is one-tenth the size of a cm.) A later German study confirmed the NIH results. This means that doctors estimate a growth rate of 3–4 mm per year when deciding upon treatment and scheduling tests and scans. An individual tumor growth rate can be calculated; there are 3 to 4 measurements made over a period of time. However, even with these, one cannot always predict the actual growth rate. Tumors often grow in steps with periods of little to no growth, followed by periods of rapid growth. Therefore, the first 2 to 3 scans should be at short intervals such as annually unless the tumors are very small.

VHLA 2013 Annual Meeting – Ann Arbor

Summary of Scientific Presentations and Patient Panels

The 2013 VHL Alliance Annual Conference was held at the Ann Arbor Regent Hotel on Saturday, September 21, 2013. The agenda included seven scientific presentations and two patient panels. The summary of presentations is posted on the VHL Alliance website at: <http://www.vhl.org/patients-caregivers/annual-patient-caregiver-meeting/annual-meeting-2013-ann-arbor/summary-of-presentations-2013>.

The presentations started with Dr. Elana Stoffel's talk, **"Monitoring the VHL Patient."** Dr. Stoffel emphasized that there is a broad spectrum of disease in VHL; the disease does not always follow what is written in books. Different types of tumors are based on VHL mutations, but there are also differences based on other gene modifiers. Environment and lifestyle factors are important; a good example is smoking. Genetic testing ensures everyone gets the care they need before manifestation of symptoms. Dr. Stoffel likes the screening grid in the *VHL Handbook*: <http://www.vhl.org/handbook>.

Genetic testing ensures everyone gets the care they need before manifestation of symptoms.

Dr. Steve Sullivan followed with a panel discussion, **"VHL and the Central Nervous System."** The panelists were Dr. Ghaus Malik and Dr. Ian McCutcheon. Both panelists agreed that the goal is as few operations as possible during the patient's life. Therefore, surgical decisions are based on symptoms while taking into account possible future problems. An example is the occurrence of multiple tumors in the brain and spinal cord. This scenario can make it difficult to decide which tumors are the sources of the symptoms. If unsure, the safest surgery is performed first. An example seen in VHL patients is the cerebellum; it is a "safe" area as one side can compensate for tissue removal from the other side. The panelists also agreed that higher location of lesions in the spine will cause more severe paralysis; a higher tumor, even though it may be smaller, is more important to remove. When possible, removal of more than one tumor in a single operation was recommended. The surgeon must always look at the risk/benefit ratio for surgery vs. not operating.

"Ophthalmology Issues in VHL" was presented by Dr. Mark Johnson. He emphasized that the retina is the only place in the body where VHL lesions can be seen directly without the use of diagnostic imaging. Ocular manifestations are often the first lesions found and lead to diagnosis. VHL is diagnosed based upon clinical appearance; there is no definitive diagnostic tool. Most lesions are in the periphery where they do not impact vision. Lesions can appear at any age and may spontaneously regress. Generally, however, the lesions keep growing slowly, eventually causing vision loss through secondary complications – retinal edema, lipid exudates, fibrosis causing retinal detachment,

and bleeding leading to neovascular glaucoma. A 2008 study by the National Eye Institute found that 38% of VHL patients had eye lesions, but 77% had 20/20 vision and only 5.7% were legally blind.

In addition to being a panelist, Dr. Ian McCutcheon gave a talk on **"Endolymphatic Sac Tumors and VHL."** This information is important for VHL patients to be aware of as these tumors are still relatively unknown even to neurosurgery residents. ELSTs are rare in the general population, but 2/3rds are not associated with VHL. Only 11–16% of VHL patients have ELSTs, and they generally occur at a younger age than in the general population. ELSTs can invade both bone and dura; MRI is good for showing the tumor, and a CT scan will show bone loss. If both VHL and tinnitus occur (92% of ELST patients experience this), the doctor should suspect an ELST.

VHL and kidney tumors were covered in two lectures. The first was a panel discussion, **"Evaluation and Treatment of Kidney Tumors,"** moderated by Dr. J. Stuart Wolf, Jr. The panelists were Dr. Elaine Caoli, Dr. Khaled Hafez, and Dr. W. Marston Linehan. The panelists agreed on the clinical approach of balance between sparing the kidneys and avoiding metastasis. The goal is to preserve the kidneys for the patient's entire life. The largest tumor generally spared is 3 cm, but the location of tumors, solid vs. non-solid (cystic), and growth rate are also important factors when deciding on whether or not to operate.



Speakers and guests from the University of Michigan

The second lecture related to kidney tumors was Dr. Linehan's **"Update on VHL Research and Clinical Trials."** There is no single kidney cancer; it is caused by different genes. On average, it takes about 20 years for a VHL kidney tumor to grow to 2 cm. There is no physiological difference between small tumors and large tumors except size; both can develop into malignant tumors. It is good to note that there is no increase in risk of adenocarcinomas of the pancreas with VHL.

The final scientific presentation was **"VHL and the Adrenals and Pancreas,"** given by Dr. Tobias Else. Dr. Else made it clear that, although pheos occur in some VHL families, all VHL patients should be checked as 20% have one. Tumors can be bilateral, occur at any age, and can be the only manifestation of VHL. There are usually

no symptoms until the tumor reaches about 2 cm. Just as in the kidneys, both NETS and pheos start as “benign” lesions; however they can result in dangerously high blood pressure. It is, therefore, very important to screen for pheos before any type of surgery.

There were two patient panel discussions following the scientific presentations at the Ann Arbor meeting. The first, “**Genetics and Childhood Issues**” was led by three genetic counselors—Victoria Raymond, Jessica Everett, and Gayun Chan-Smutko. The panelists were Danielle K., Jeff Y., and Andrea B. The panelists discussed how to inform children about VHL. Children can have a range of reactions from anger at the parent who passed along VHL to anxiety about the uncertainty of symptoms associated with VHL. The moderators recommended using the *VHL Handbook Kid’s Edition* (<http://www.vhl.org/handbook>) to help children develop a vocabulary about VHL. There was also a dialog of when parents should talk to their children with VHL about the child’s plans to have a family. They concluded that each child is different, however in their experience, early teens would be appropriate. A genetic counselor can be helpful in explaining options to teens and young adults. Finally, the panelists spoke about how they decided with whom and when to share their VHL diagnosis. It was generally

agreed to share only with those you are close to and when the time felt right.

The second VHL patient panel discussion was “**The Emotional Roller Coaster**,” moderated by Wendy Uhlmann. The panelists were Amber M., Shawn M., and Molly O. The panelists all agreed that VHL follows an emotional up and down course. Shawn views VHL as a similar type of challenge to the marathons he runs as a “VHL Warrior.” Events causing stress include: going for yearly scans and waiting for the results; changing doctors; the decision on whether or not to have children; dealing with bad news from test results or unexpected recommendations; and preparing for surgery. Strategies that have worked for the panelists are: going to a doctor who is an expert and with whom you have a positive relationship; researching VHL and getting multiple opinions when there are any questions about diagnosis or treatment; making the present day as enjoyable as possible—for example, the annual trip for scans and tests can be a “fun” day by eating out or shopping and taking control of your decisions and how you live—not letting VHL control your life. Involvement with the VHL Alliance, including attendance at the annual meetings, provides in-person education and the opportunity to meet others with VHL.

HAPPENINGS AT THE VHL ALLIANCE

Fundraising Events

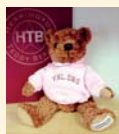
On August 24, 2013, the Andjus family of Massachusetts held their second annual **Cornhole for a Cause**, which was once again a huge success. There were over 75 people at the event and 20 teams competing. A total of \$8,343 was raised.

Kelly Mackesey of Maryland hosted a **Silent Auction with Raffles** and raised over \$6,500 in October, 2013.

On September 22nd, **Team VHL** was launched in Ann Arbor, Michigan. The FUNdraiser raised almost \$20,000 for VHL research, education, and support! To see more pictures, go to our website: <http://www.vhl.org/wordpress/ways-to-help/fundraising-ideas/join-team-vhl>. **Join Team VHL!**



Louella Rosie Jones of NY, dear friend of the late Altheada Johnson, also hosted a silent auction, selling her Stella & Dot Jewelry to benefit **Light and Hope for VHL** on December 4th. “Our beloved Altheada LaVerne was smiling down and I think I did her proud.” \$550 was raised.



On January 30, 2014, Manuel Greco will be hosting a **New York City Fundraiser to benefit VHL** at Il Gattopardo Restaurant. <http://www.vhl.org/january-2014-nyc-fundraiser>

You Can Make a Difference

Have you really wanted to make a difference in your life to help yourself, family members, or friends as well as your cause? VHL Awareness, Education, and Funding for VHL Research are the top goals of the VHL Alliance.

Everyone can help! There are many things you can do to raise funds to support VHLA. You can go to our website: <http://www.vhl.org/ways-to-help/fundraising-ideas> or call the office at 1-800-767-4845 ext. 4.

Here is a very simple way to help. Sign up for your own fundraising page! The funds will go directly to the VHL Alliance It’s easy to do. **Sign up, Set up, Send e-mails.**

For step-by-step instructions, go to the VHLA website at <http://www.vhl.org/library/How-to-do-Online-Fundraising-for-FirstGiving1.pdf> or call the office at 1-800-767-4845 ext. 4.



Chapter Meet and Greets

On December 13, 2013, the **New York Chapter** held a casual end-of-year **Meet and Greet** to get to know other VHL patients and families. They gathered at Vapiano’s restaurant for drinks and hors d’oeuvres.

On January 17, 2014, the Canadian VHL Alliance had their **Toronto Meet and Greet** in Mississauga, Ontario. www.vhl.org/canada

Donations are seen on the printed version only.

Your help is greatly needed

DONATE

We also need volunteers. Call 1-800-767-4845 ext. 4

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October 23–25, 2014

**International VHL Medical
Symposium 2014, Madrid**

<http://www.vhl.org/symposium2014>

Saturday, October 18, 2014

2014 VHLA Annual Meeting, Rockville, MD

<http://www.vhl.org/meeting/annual-meeting-2014>

3rd Annual Fundraising Dinner & Auction

honoring W. Marston Linehan

<http://www.vhl.org/meeting/dinner2014>

Sunday, October 19, 2014

Team VHL Fundraiser

Details to follow

See website for more details or call the office at 1-800-767-4845 ext. 4