Join us for VHL Awareness Month

By Heidi A. Leone, Director of Advancement

Recently, the VHLA Board of Directors decided to update my title to Director of Advancement. I am excited about this change as I really think it encompasses more of what I do at the VHL Alliance. I wear many hats: some of my responsibilities include fundraising, marketing, awareness, as well as coordinating events and volunteers. Because I am working on these things on a daily basis, each day and every month is VHL Awareness Month to me—I am always striving to get VHL and VHLA promoted and recognized!

In order to emphasize the importance of VHL awareness, May has been designated as VHL Awareness Month, internationally! We hope that all of you help us bring awareness to this disease by participating this May. There are fun and creative activities that are already being planned, and we encourage you to add an activity near you if we do not already have one.

Several different groups of people are meeting for cocktails and appetizers at homes or restaurants; one family is selling pies from a local bakery—where a portion of the proceeds go to VHLA. Other people prefer to do things virtually and have created their own individual fundraising page at grouprev.com/vhlliance. Another family is doing a costume walk where everyone dresses up in a crazy costume and walks around the town pond! Sunday, May 8th is Mother's Day. Team VHL is hosting a Mother's Day run at Ventura State Beach in California. What could you do to honor your mother while promoting VHL awareness? How about a friend-raiser or fund-raiser at your children's school or at your church?

Some of the most effective awareness events we have found are regional gatherings, auctions, sports competitions, fashion shows, and wine tastings, to name a few. There are more ideas and ways to get started in our Awareness & Fundraising Toolkit that can be downloaded for free at vhl.org/get-involved/fundraise. Would you be willing to put something together? Give me a call and we can brainstorm together!

In honor of VHL Awareness month, VHLA has put together various materials to help you spread the word. Cards to pass out, profile pictures to post on social media, and blurbs to put on your email signatures are all available for your use. Also, please follow us: Twitter, Facebook, and Instagram will all be ramped up for May!

Each person who becomes aware of VHL is a potential friend of VHLA. Those donations are what are putting us closer and closer to a cure for VHL and other forms of cancer as well as allowing us to continue to support and educate the VHL community. Thank you for your help!

New Screening Test for VHL Patients?

Anouk N.A. van der Horst-Schrivers, MD, PhD, Endocrinologist, University Medical Center Groningen, Groningen, The Netherlands. 2016 recipient of the VHLA-funded pilot grant

Pheochromocytomas are tumors arising from the marrow of the adrenal gland (adrenal medulla). In von Hippel Lindau (VHL) disease, they occur in about 18–26% of patients. Pheochromocytomas give rise to symptoms like headaches, sweating, palpitations and high blood pressure. These symptoms are not caused by the tumor itself, but caused by the hormones that are produced and secreted by the tumor: epinephrine and norepinephrine. Together they are called catecholamines. Sometimes, these catecholamines are released in large amounts, which not only leads to headaches, but also to very dangerous conditions such as a heart attack or an infarction of the brain. Therefore, it is important to screen for these pheochromocytomas.

Screening for pheochromocytoma consists of demonstrating the excess of catecholamines. Since these catecholamines are not released...
continuous but only during crises, the epinephrine and norepinephrine values in blood can be normal between crises. In medical terms this is what we call a “false negative” test result (i.e. the test results are normal, although a pheochromocytoma is present). Hence, we measure the factors that are formed when catecholamines are metabolized: these factors (metanephrines) are also elevated in between attacks. Epinephrine is metabolized into metanephrine and norepinephrine is metabolized into normetanephrine. By measuring the metanephrines preferably in blood (or in 24-hour urine collection), almost all pheochromocytomas are detected.

Patients with VHL are annually screened for pheochromocytomas using a blood sample (or 24-hour urine collection). This starts at an early age, since a pheochromocytoma can develop in early childhood. The VHL Alliance, in conjunction, with their medical advisory board recommends annual screening from the age of 5 years.

The disadvantage of screening with a blood sample for a pheochromocytoma is not the false negative tests, but the false positive tests. In such a circumstance, the test result is positive, but a pheochromocytoma is not present. This is caused by the fact that norepinephrine is not only produced and secreted by the adrenal gland but also by the nervous system, as a kind of messenger.

When a person is in an upright or seated position, the nervous system is activated and the norepinephrine production of the nervous system goes up, and, thus, also the normetanephrine production. This can lead to a false positive result. To avoid this, patients must rest for 20–30 minutes in a supine position before taking a blood sample.

Saliva is in many ways equivalent to blood; several hormones and factors, that are present in the blood are also present in saliva, although in a lower concentration, such as metanephrines. Saliva is very easy to collect using a cotton pad, even at home. You just keep the cotton pad a few minutes in your mouth, put it in a tube and send it to the hospital, a few weeks before the medical appointment.

Measuring metanephrines in saliva could be less cumbersome and more patient friendly, especially in children, since blood draws are avoided.

We recently published a pilot study (a small study)¹ to see if metanephrines can be measured in the saliva of a healthy person. This study shows that this is indeed possible. The study also showed that normetanephrine levels in saliva were lower after the healthy person had been in a supine position for 30 minutes, and that it is better to collect the saliva more than 30 minutes after

With funding from the VHL Alliance, we are now testing the feasibility of an easier method of detecting pheochromocytoma.


Eliminating Isolation with a Rare Disease

Written by Matthew Carli

Receiving a diagnosis for any serious disease, from cancer to diabetes, can be devastating. As patients begin to adjust to their new reality, they often find it helpful to reach out to others with the same disease. By sharing advice and experiences unique to their condition, these patient communities provide a great deal of comfort. For most people, finding others living with the same disease is easy, but for those who suffer from a rare disease it can be much more difficult.

A disease or disorder is defined as rare in America when it affects fewer than 200,000 Americans at any given time¹. Because so few people are touched by these conditions, finding fellow patients locally is often impossible and in-person support groups may not exist. Unable to communicate with others going through the same experience, rare disease patients can begin to feel as though they alone are dealing with their condition.

Most people understand that isolation is not healthy. Studies have shown that social isolation negatively affects health in a variety of ways². Ideally, patients diagnosed with a rare disease will have family and friends to lean on, which acts as a strong buffer against isolation³. When current relationships do not meet the needs of their particular situation, those living with a rare disease must seek out new contacts themselves.

The bright side to this situation is that in today’s world it has never been easier to communicate with others. The internet has revolutionized how people interact and allows people around the globe to reach out to each other with ease. A quick search can reveal a website dedicated to a rare disease. Conversations
in a Facebook group (or other social networking venues) allow people separated by an ocean to talk almost as if they were face to face. However, even these online support groups and patient organizations are sometimes sparsely populated with few active members.

The solution to this problem is to become active. The standard recommendation in any case of social isolation is to consciously increase interaction with the outside world4. In the case of rare disease patients, this engagement is even more crucial. Because there are so few patients for any given rare disease, it is more important for each person to actively participate in the community. For diseases where patients have already organized a group, contributing can be as simple as sharing a personal story and sparking conversation, or volunteering time to handling administrative tasks the group needs done. The key is to be active because that activity will encourage others.

Patients touched by rare diseases all share common problems and goals, from finding the right doctor to spurring on more research for rare diseases. Getting involved with your rare disease community is a great way to combat the isolation of rare disease. NORD’s motto says it well: “Alone we are rare. Together we are strong.”

REFERENCES

Fun and Easy Ways to Host a VHL Fundraising Event

By April R.

For my birthday each year I always get a group of friends together and find fun ways to celebrate another year. After all, let’s be honest, another year conquering VHL is another year to celebrate! I combine this celebration with a way to raise money that will help find a cure for this disabling disease.

Over the last several years, I have hosted a variety of events and donated the proceeds of these events to the VHL Alliance. Painting classes have been a fun way to get a group of us together and paint for a cause. I have partnered up with a local artist that has been willing to donate the proceeds of our painting class to VHLA; we add a little extra cost on to the normal class charge to contribute even more money! My friends and family really enjoy coming together for this social event while, at the same time, creating a painting to take home AND making a financial contribution to help with the elimination of this disease. I would encourage all of you to locate a craft store, or painting class or hobby in your hometown. Find a business owner that is willing to partner with you by donating their proceeds for a cause. Then get a group of people together for a fun night out, charge extra, and donate to VHLA.

Another fund-raising event I have done is hosting a barbeque that includes games and food. Guests can make donations and enjoy a time of fellowship and fun! Hosting a garage sale is another way I have raised money. Invite friends and family to do some spring cleaning, collect items, advertise this sale, and donate the proceeds to our cause.

Churches in communities are often looking for service projects and may wish to partner with you to create additional fundraising ideas and opportunities. VHLA has sent us T-shirts, bracelets, and pins to sell along with brochures to distribute at the events in order to educate others about this disease.

If hosting an event seems to daunting to you, you can always create a personal fundraising page at grouprev.com/vhlalliance. This can be done on its own or in conjunction with an event, so family and friends near and far have an opportunity to support your cause.

The only way we are going to find a cure for VHL is to raise money for research, and we are the best advocates for our future and our children’s future.

Do you have VHL? Have you registered with the Databank?

If so, thank you! That is a great first step to helping researchers work on a cure for VHL. But it is only the first step. With every scan, MRI, and doctor’s appointment, your medical history changes and your survey needs to be updated. Please update your surveys and upload or send us your scans today!!

For any questions, write to suzanne.nylander@vhl.org.
Off-Label Use of Prescription Medications

All medications, both prescription and “over-the-counter,” sold in the United States must first be approved by the Food and Drug Administration (FDA). The approval has two parts: the drug must be safe, and it must be effective for a specific use. “Off-label drug use refers to the practice of prescribing a drug for a different purpose than what the FDA approved. This practice is called “off-label” because the drug is being used in a way not described on its package insert (label).”1

Once the FDA approves a drug, doctors can prescribe it for any purpose they think makes sense for the patient. Off-label uses may include using an approved drug for a different type of cancer than the one it is approved to treat, at a different dose or frequency, and to treat a child when it is approved to treat adults.2

The FDA is aware that off-label prescribing of drugs is very common. “In 2001, 150 million off-label prescriptions were written—21% of all prescriptions for 160 common medications in the U.S. ...Up to 75% of drug use in cancer care and about 90% of drug use in rare diseases is off-label.”3 The FDA states that “good medical practice and the best interests of the patient require that physicians use legally available drugs, biologics and other devices according to their best knowledge and judgement. If physicians use a product for an indication not in the approved labeling, they have the responsibility to be well informed about the product, to base its use on firm scientific rationale and on sound medical evidence, and to maintain records of the product’s use and effects.”4 Although the FDA would like physicians to note off-label drug use, the US “does not require physicians to record the purposes for which they prescribe drugs...on each prescription...[including] the patient’s principal diagnostic and symptom codes, gender, and age... This prescribing data would identify the therapeutic goals of off-label uses and the affected populations... It would reveal when there was a need to sponsor drug education regarding particular drugs or medical problems.”5

Pharmaceutical companies are not allowed to advertise their drugs for off-label use, even if more prescriptions are written off-label than for the approved use. Physicians, however, are free to discuss off-label uses for drugs, including articles written for publication and at physician continuing education lectures and at industry-sponsored presentations. This has allowed off-label use of drugs to become standard clinical practice. For example, aspirin was an accepted drug prior to the creation of the FDA in 1938. Aspirin has been approved by the FDA for numerous conditions such as pain, fever, and as a prophylactic (preventive) treatment for heart disease. However, aspirin prophylaxis for diabetic patients is not an approved use, “yet guidelines recommend its use in these patients. Therefore, aspirin prophylaxis for coronary disease in high-risk patients is an off-label use.”6 Another area where off-label use is common is in psychiatry. “Patients with psychiatric disorders are often excluded from clinical trials...There is often crossover in symptoms from disease state to disease state, which has lead physicians to use psychiatric medications approved for one psychiatric condition for additional unproved indications...[It is] estimated that the cost of off-label antipsychotic drug use in 2008 was $6.0 billion.”7

Because off-label prescribing of drugs is so common, “doctors will often prescribe a drug off-label without realizing it is not approved for that use...How can you protect yourself from...potential hazards of off-label drug use?...When your doctor prescribes a drug, ask if it is an approved use. If he or she doesn’t know, ask your pharmacist. Check for yourself. Go to DailyMed (dailymed.nlm.nih.gov) and search for the drug. Then click on the tab for “Indications & Usage” to see if your condition is listed. If it is an off-label use, ask your doctor if...trials show significant improvement for...your condition...Find out if your health insurer covers payment for the off-label use. Some may require evidence of effectiveness or failure with conventional treatments, especially if the drug is expensive.”8 You will need to be proactive and ask your doctor if a drug is being prescribed for you off-label as there is currently no legal requirement for your doctor to obtain your informed consent that you will be taking a drug for an unapproved indication.

With off-label use of drugs so common, you may ask why the drug manufacturers do not conduct clinical trials and obtain approval for additional medical conditions that respond to their drugs. The simple answer is that there is no financial incentive. “Obtaining a new FDA approval for a medication can be costly and time-consuming. To add additional indications to an already approved medication requires the proprietor to file a supplemental drug application and, even if eventually approved, revenues for the new indication may not offset the expense and effort of obtaining approval.”9

Health insurance companies are concerned about off-label drugs “because the use of off-label therapies continues to climb, contributing to the escalating cost of health care... U.S. prescription sales amounted to $291 billion in 2008...That would put off-label usage at nearly $60 billion.”10 Private health insurance frequently bases decisions on coverage on federal Medicare rules. Medicare expanded the list sources (compendia) for approved drugs in 2009 to include many off-label indications. There is agreement that these lists are subjective, but they do provide a third-party opinion when an insurer is deciding on coverage for an off-label prescription.

Your knowledge, as a patient, of the pros and cons of off-label prescribing will help you better understand your medications as well as assist you in any discussions about coverage with your health insurance company.
Annual Mom’s Day Run
On May 8th, in Ventura Beach, CA, Team VHL will again host the Mother’s Day fun run/walk along the beach. Join us with your mom, in honor of your mom, or in memory of your mom.
Emily B, VHL Warrior and former contestant on NBC’s The Voice, will again perform!
tinyurl.com/2016MomsDayRun

Recent
Wisconsin Chapter
Wisconsin Chapter Leader, Sandy, recently partnered with the Elegant Farmer, a local bakery, to do a fundraiser in her area. With the help of family members, friends, and a VHL patient, sold over 125 pies, with a portion of each sale going to VHLA!! Along with each pie sold, she shared information about VHL and VHLA. Talk about great awareness! Thank you Sandy!

Minnesota Chapter
Over the weekend of February 20th, the VHLA Minnesota Chapter hosted a get together. This was an informal gathering to bring together VHL patients and families to share knowledge, develop connections for future support, understand the latest screenings and treatment options, and to make everyone aware of all resources related to VHL. 24 people attended. The conversations were great!

RUNdezvous 5K Couples Run in California
On February 13th in Newbury Park, CA, Team VHL Captain Suellen led a couples run to benefit the VHL Alliance.
This is the 2nd Annual run on Valentine’s Day weekend. Here is an article from the Ventura County Star:
tinyurl.com/RUNdevouz2016

Create your own Fundraising Page
It’s easy to set up your own fundraising page for your May Awareness activity. Go to grouprev.com/vhlalliance and click on “Create Your Own Page”, put in your own title, story and your goal. GroupRev makes it easy to follow along. Then e-mail your page to friends and family or post to Facebook and Twitter.

Team VHL Baltimore
Join Team VHL at the event that many are calling one of the best races on the East Coast. There are five distances to choose from. You won’t want to miss the 15th Anniversary Baltimore Running Festival with some special added features to celebrate this historic benchmark.
At the time of publication, there are already 17 people signed up to run!
For more details or to donate, go to grouprev.com/teamvhlbaltimore or write to team_vhl@vhl.org

VHL Insurance Denial Letters Needed
Have you ever had a health insurance denial for VHL screening tests or treatments? If so, please let VHLA know. If you have a copy of a denial letter, we would appreciate that, too. VHLA is gearing up to advocate on the state and federal level for improved health insurance coverage. Examples will be helpful Please email this information to suzanne.nylander@vhl.org or mail a copy to the VHLA office.
Thanks so much for your help with this effort!

References
1,2 National Cancer Institute; “Off-Label Drug Use in Cancer Treatment”; cancer.gov, accessed 3/25/15
4 FDA Regulatory Information; “Off-Label and Investigational Use of Marketed Drugs, Biologics, and Medical Devices—Information Sheet”; fda.gov, accessed 3/25/15
5 Marc Rodwin; “Managing Off-Label Drug Use”; Health Affairs Blog, December 17, 2013
8 Consumer Reports; “Off-label drug prescribing: what does it mean for you?”; consumerreports.org, December 2012
10 Lola Butcher; “When Should Insurers Cover Off-label Drug Usage?; Managed Care, May 2009
2016 VHL FAMILY ANNUAL WEEKEND
Los Angeles, CA and surrounding area

• Fundraising Dinner & Auction

HONORING
Michael G. Gorin, MD, PhD
Harold and Pauline Price Professor of Ophthalmology, Chief of the Division of Retinal Disorders and Ophthalmic Genetics, Department of Ophthalmology UCLA David Geffen School of Medicine

Suellen Lindquist
VHL patient, caregiver, Team VHL Captain, enthusiastic runner and organizer of running events and T-shirt sales to benefit VHLA, inspiration to many.

Friday, September 23
Manhattan Beach Marriott, 1400 Parkview Avenue, Manhattan Beach, CA 90266

More details, tickets, and sponsorship opportunities. See tinyurl.com/VHLweekend2016

• Annual Family Meeting

Saturday, September 24
RPB Auditorium, Jules Stein Eye Institute at UCLA, 100 Stein Plaza, Westwood, CA 90025

tinyurl.com/VHLweekend2016

AGENDA TOPICS - Speakers include doctors associated with UCLA and others associated with the VHL Alliance

- Genetic Issues and Challenges in the Coordination of Care
- The Emotional Roller Coaster of VHL
- Treating the Kidney: Surgery vs. Radio Ablation
- Retina Lesion Detection and Treatment
- Adrenal issues and surgery
- Neurosurgery: When and How to Operate
- Living with Chronic Pain
- Childbearing Options
- Patients and Doctor Learning Together

• 2016 Team VHL 5K Run/Walk

Sunday, September 25
Wood Chip Trail, Manhattan Beach, CA
(walking distance from the Manhattan Beach Marriott)

DETAILS/REGISTRATION: tinyurl.com/teamvhl2016

• Concert by Emily B and Friends

Sunday, September 25, 1:00 PM

More information to come

CHECK BACK AT: tinyurl.com/VHLweekend2016

LODGING
Manhattan Beach Marriott
1400 Parkview Avenue, Manhattan Beach, CA 90266
For reservations, call 800.228.9290 (group code: VHLVHLA)
or register on-line at cwp.marriott.com/laxmn/vhlalliance/

You must book by 08.22.2016 to receive VHLA's special rate!

If you'd like to donate an item for the auction, please contact us at 617.277.5667 ext. 4.
Our Sincerest Thank Yous

Donations are seen on the printed version only.

Your help is greatly needed

DONATE
Join us for the VHL Alliance
Annual Family Weekend

September 23–25, 2016
Los Angeles, CA

Fundraising Dinner & Auction
Annual Family Meeting
2016 Team VHL 5K Run/Walk
Concert by Emily B & Friends

For details on all events, lodging, and registration, go to tinyurl.com/VHLweekend2016
Call the office with any questions: 617.277.5667 x4 or e-mail office@vhl.org