

ANNUAL  
REPORT

## 2015-2016



The quarterly newsletter of the von Hippel-Lindau Alliance

## Think Big and Be Noble

by Doug Karle, VHLA Board Member



Eight strokes caused during a pheo crisis, two pheochromocytoma, one bilateral adrenalectomy, two heman-gioblastomas, one paraganglioma and a working diagnosis of “likely metastatic pheochromocytoma”... and I feel lucky.

A father who had VHL and died too young, one sister who had a pheo removed earlier this year, and one child who inherited this genetic nightmare ... yet I am thankful.

As sneaky, random, and punishing as VHL can be, all of us involved with the VHL Alliance make it an invaluable gift. We get to see the world as almost no one else does. So many people give time, talent, and treasure to hopefully prevent others from going through what we have and to hopefully prevent their kids from ever knowing VHL.

Beyond benefiting VHL patients, studying VHL is a way to help prevent the death of 1,500 people every day at the hands of cancer. Not simply a cure for VHL, but our research is a significant contributor in the cure for many cancers. Those of us associated with VHL have been given an opportunity to do something great. How can the approximately 10,000 people with VHL in the United States, and all those connected to them, be a part of this? Consider the following:

- Research related to VHL has resulted in eight cancer drugs currently on the market.
- The pathway of the VHL gene controls the major feeding pipeline of many cancers. There is a good chance that on the third allele of the third chromosome lies research and effort that will lead to a cure outside of VHL.
- Data from the VHL Patient Databank ([vhl.org/databank](http://vhl.org/databank)) will help change how we think about VHL and cancer in general.

There is a debate as to whether VHL is a cancer or not. For me, it is not a debate; I can only describe it as endless cancer. We do not get to count to five years, looks at the statistics and declare, “I am in the clear.” VHL does not stop. It never rests—its footpath both unplanned and harsh. So what do I do with this reality? We only have one choice ... live it and own it with a rebellious spirit.

Life for everyone who has VHL is extremely difficult at best, and being a parent of a child with VHL can be excruciating. The tears are never far below the surface, even during happy times.

It happens casually and without noticeable triggers. A few days ago, I was watching my son play with his best friend. I was brushed back to when I was seven and recalled what I was feeling, loving, and pursuing in a similar carefree moment. Knowing what is brewing inside of him was unbearable; I had to leave the room to wipe away my tears. I want to be there for him as he grows. I want to be able to connect as no other person will be able to do and to offer my perspective, compassion, and love. With my working diagnosis, this may or may not happen. I do not know how he will react in his teen years when he will most likely develop his first tumor and, most importantly, I want this to just go away for him.

I am intensely aware of our priceless time we have on earth. We have one life with no guarantees and no do-overs. One thing that I know to be true through is that we get more than we give, so let's all **think big and be noble**. Let's work to save the lives of adults and children and to give them the gift of time; time with their parents, their children, their loved ones, and the opportunity to positively impact with everyone they meet. Curing VHL is not my only goal, I see it also as the avenue to a greater outcome.

**Unprecedented Matching Gift Opportunity for VHLA!**

See page 4 for details

# Welcome Andrea Berkemeier!



The VHL Alliance is thrilled to introduce our newest employee, Andrea Berkemeier. Andrea joined VHLA in September as Director of Engagement and Outreach. Her portfolio includes patient support, working with the VHL Clinical Care Centers, and team leader for the Patient Databank. Andrea is already personally reaching out to many VHL stakeholders, initiating teen and young adult groups, and working on creating new CCCs.

Andrea's interest in VHL began at age 12 when she was diagnosed with VHL. This spurred her interest in using medicine as a means of improving the lives of others. She pursued her BSE in Biomedical Engineering at the University of Michigan and earned her MA in Bioethics & Society from King's College in London. For her Master's dissertation, she examined how VHL patients navigate ethical considerations pertaining to family planning.

## VHL Teens at the Annual Family Weekend

by Melissa H., VHL Patient

This year the VHL Alliance Annual Family Weekend was held in Los Angeles, California. Because I was recently diagnosed with VHL, it was the first year that I went to the meeting. My name is Melissa and I am 16 years old. Because I am still a teenager, I got to take part in the teen activities. This was the first time that VHLA ever held a teen event. There was an amazing scavenger hunt, up and down the Santa Monica Pier. The items on the hunt were things such as the end of Route 66 and Muscle Beach! This was definitely something I would recommend to anyone. The people there were so nice. It felt good to meet others battling the same disease as I am.

The Annual Meeting included doctors from across the country who came to talk about their specialties and how

they treat VHL. The presentations were very informative, and they gave new insight into research being done towards better treatment and eventually finding a cure. After all the presentations, people broke up into two groups, patients and caregivers, and we just talked about our struggles. This was such a great experience for me; it definitely helped get some stress out as well as a few things off of my chest. Because many of them have been through struggles that come with VHL, they have a lot of insight into how to make you feel better about VHL and to get through it. This is something that I really appreciate.

Next year I am planning on going again. I really enjoyed the trip, and I hope that other people can experience this the same way I have. I can't wait to meet new people and more VHL Warriors. **After all, we have to stick together!**

## The VHL Alliance Helps Us Connect

Ed W., VHL Patient and Chapter Leader

The VHL Alliance has helped many of us in the VHL community connect in ways that otherwise might not have been attainable. Many of us have connected through the VHLA Facebook page (@VHLAlliance) which has led to in-person meetings, emails, as well as telephone conversations.

I have always wanted to be a voice for VHL. A few months ago, I had the opportunity to share my VHL story with a Pennsylvania state senator whom I met at a local parade. Going to the parade that day and posting my experience on the VHL Facebook page gave me the opportunity to make my voice heard for the first time. As it turns out, this particular Senator, Sean Wiley, has a strong interest

in Rare Diseases. I was able to share with him some specific information about VHL, including leaving him a copy of the VHL Handbook for his reference. Our hope is that he can advocate for support and awareness of VHL. I am grateful that we have the support from the VHL Alliance and that I have the opportunity to be a Chapter Leader in my region—to be the eyes and ears of the VHLA in our local communities.

My desire is to represent fellow VHL Warriors; to bring awareness, and to teach other about our disease. It has been a joy learning more about VHL through the Alliance and my work as a Chapter Leader. Other Chapter Leaders are needed—please join me in this important role by contacting Heidi.Leone@vhl.org.

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# A TraVHLers Guide

by Wes S., VHL Caregiver and Chapter Leader

When my wife was diagnosed with VHL in 2005, it was hard to see how anything good could come from it. The next ten years would be an endless stream of appointments and scans all too familiar to those in this community. But believe it or not, there is a better side to this syndrome than just waiting to be told you will need surgery.



Rather than become denizens of the same old places to which VHL has relegated us, we wondered if we could explore beyond, to experience something related to this disease that might even resemble, dare I say it...fun.

So, we decided to travel, not to another meeting with the neurosurgeon or retinal specialist, but to Boston for the International VHL Medical Symposium. Boston is a place we have always wanted to visit.

We met some of the brightest minds in medicine and had the chance to meet other VHL Warriors from all over the world, sharing fears, stories, and scars. We heard presentations from caregivers, teens, and young adults in addition to the myriad medical professionals who have come together for this common goal.

When we were not at the symposium, we were taking in all that Boston has to offer. For the first time since being diagnosed, we did something relating to VHL that was, dare I say it ... fun.

So when the next opportunity came up, the Annual VHL Family Meeting, we did not hesitate; this time, it was Los Angeles, California. We had never been there either.

The Family Meeting is not a Medical Symposium but a casual conference where patients and caregivers can hear from and talk directly to doctors and researchers about anything they please. We found the programs to be very enjoyable and informative. Small groups and breakout sessions for patients and caregivers were also a treat, allowing us to get to know other families in a more meaningful way.

It was our first time to the West Coast as a family and we made the most of the trip. We hung out with our kids and watched as they left our sides to go hang out with other kids in families with VHL.

I felt happy my wife was getting to experience VHL, not as a wounded patient in need of surgery, but as an empowered Warrior surrounded by other Warriors. She collected so many contacts, met so many new people and above all, made many new friends that we are sure will be there down the road.

We encourage you, if you are living with VHL, to do all you can to experience it in a new way, bereft of hospitals and waiting rooms. Experience VHL as part of a community, in a new place with new friends. It may actually be ... fun.

## Steps Toward a Cure

by Eric Jonasch, MD, VHLA Board Member

Advancing our scientific understanding of VHL disease is one of the top priorities for VHLA. To that end, we implement a research grant program that is rapidly attracting some of the top names in the VHL research world. The goal of the grant program is to fund projects that will: 1) increase our understanding of how VHL affects the cell; 2) permit the creation of model systems that better mimic the organ involvement of VHL; and 3) will develop improved screening and treatment approaches for people living with VHL. These grants come in two sizes: a one-year \$25,000 pilot grant and a two-year \$100,000 research grant.

VHLA has funded several exciting new projects in the past few years. Dr. Horst-Shrivers from the University Medical Center in Groningen received a pilot grant award. With the hope of replacing the current blood or urine tests for detecting pheochromocytomas, she is testing whether the hormones released can be reliably measured in saliva. This approach, if successful, will clearly make the lives of individuals living with VHL a lot easier. Dr. Othon Iliopoulos, Associate Professor at the Massachusetts General Hospital in Boston, is using VHL zebrafish to screen for compounds that may alter the effect of VHL mutations. Zebrafish with the VHL gene deleted display

a number of VHL lesions similar to those seen in people. Since these tiny fish are relatively transparent, you can actually "see" the effect that potential drugs have on the VHL manifestations. Professor Ian Frew at the University of Zurich is developing a model of kidney cancer in mice, a task that has been quite difficult to achieve. Dr. Frew will be able to use this model to test new drugs that can treat kidney cancer.

In 2016, we have two new awardees: **Dr. Raymond Kim** from the University of Toronto and **Dr. Michael Gorin** from the University of California. Dr. Kim will head the VHL-IT Sharing International Consortium (VISION) with the goal of developing a more efficient approach to collect information on VHL mutations and the way that VHL manifests in these individuals. Dr. Gorin will work on developing two new models to study VHL retinal lesions, one model will use pluripotent stem cells; the other model is a VHL knockout mouse.

We anticipate that these talented investigators will help move the field of VHL research forward substantially in the next few years. This exciting research is supported by donations from the VHL community. With your support, we look forward to funding additional groundbreaking research projects in the years to come.

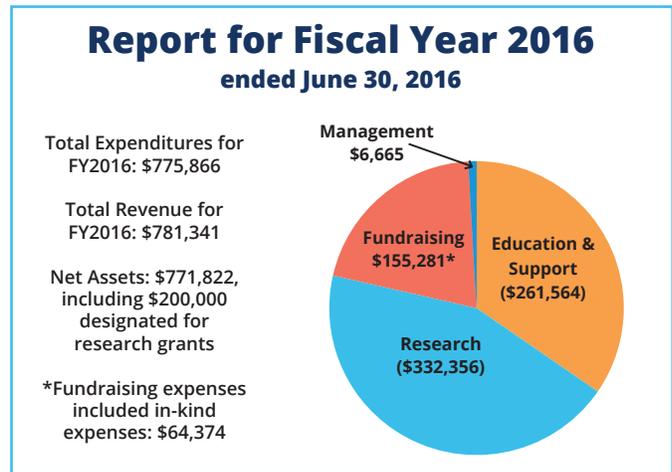
# FY16 Financial Report

*by Andrea Rafael, VHLA Treasurer*

The work of the VHL Alliance is difficult. Together, with our hard working staff, we need to continue to expand ways to support our stakeholders, further educate patients and healthcare professionals, alike, raise awareness of the involvement of the VHL gene pathway in many forms of cancer, and continue to fund important research leading to a cure.

It takes teamwork to live our mission and to ultimately reach our dream of finding a cure for VHL. We are all part of that team! Each year we ask that all those directly or indirectly impacted by VHL to financially contribute to the VHL Alliance. This year we ask that you dig as deep as possible. We at the VHL Alliance know that that amount is going to be different for each individual, but we also know that together we can reach our goal of raising \$275,000 by the end of the year and \$825,000 by June 30, 2017, the end of our fiscal year. We owe this to ourselves, our children, their children, and generations to come. Together, let us each commit to giving forward in a meaningful way!

Thanks to your contributions, we raised over \$775,000 during our 2015 fiscal year. Research was the greatest expenditure; research to find the cure. Together we can exceed last year's success by further enabling our capacity to achieve our mission: "VHLA is dedicated to research, education, and support to improve diagnosis, treatment, and quality of life for those affected by VHL".



## Help Find a Cure for VHL!

### Every New Dollar Matched!

The VHL Alliance is thrilled to announce that several generous donors have provided \$62,500 towards an **unprecedented matching gift opportunity for VHLA**.

All **new** gifts, or gifts from donors who have not given in at least three years, as well as gifts that are **increased** over previous donations, will be matched dollar-per-dollar.

When the Challenge Grant is met, **\$125,000** will go to VHLA, supporting research, education, and support. Please help us make the most of this generous offer, between **now and December 31**, as funds remain available.

Thank you to the following families for taking part in this challenge: Bakke, Buckley, Canning, Greene, Horwitz, Johnson, and anonymous.

***Thank you!***

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

*For more information, contact the VHLA staff at [info@vhl.org](mailto:info@vhl.org) or 617.277.5667 x4.*