

von Hippel-Lindau Research Campaign

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VHL Family Alliance: A Source of Hope and A Place for Action

by Michelle Cieslak, Treasurer of the VHL Board of Directors

Like any young mother, I was excited by the birth of our son. He was perfect – ten fingers and ten toes! With the arrival of a healthy child, we counted our blessings.

At age 12, things changed! Our “healthy” preadolescent began complaining about his vision. The results of a routine eye exam prompted a visit to a retina specialist and a genetic counselor. In the end, we were told our son had a rare genetic disorder called von Hippel-Lindau (VHL).

With no family history, my husband and I had never heard of VHL. Fortunately, an Internet search connected us to the VHL Family Alliance (www.vhl.org). During our Internet crash course, we learned that VHL is caused by a genetic mutation. As well as providing critical knowledge, the VHLFA gave us *hope*. *Hope* in the form of:

- Community, a place of comfort from others who understand.
- Medical and Research Advisory Boards comprised of world renowned VHL experts.
- Clinical Care Centers where our son receives expert care.
- Guidelines for routine screening as a means of monitoring tumor growth.
- Medical research which has led to the development of drugs being used to treat kidney cancer and soon to be tested in VHL patients.

**Please join me in supporting
the VHL Family Alliance.**

My *hope* has prompted us to take *action*! Today, I am actively involved in VHLFA. As Treasurer of the Board of Directors and Chair of the VHLFA Fundraising Committee, I understand how actions substantiate hope.



The Cieslak family

The *hope* continues. At present, the VHLFA is in the process of launching an International Patient Registry. This registry is designed to help researchers better understand the manifestations of VHL. It will also provide necessary data for our new Clinical Trials Task Force charged with developing clinical trial protocols.

Thanks to scientific research on the VHL gene, pharmaceutical companies have been creating new therapies (called antigenic inhibitors) to prevent the growth of blood vessels – the underlying cause of tumor development. Some of these drugs are already on the market for kidney cancer and have implications for VHL tumors as well as other forms of cancer. The VHLFA Clinical Trials Task Force is exploring ways in which our patient population can benefit from these therapies.

I hope that you will join me in taking *action*!

Volunteer – please consider joining a committee; **Donate** – every dollar counts; **Fundraise** in support of medical research – VHL research benefits all cancer research.

Our action today will fuel the hopes and dreams of tomorrow!

Quotes from Great Nonprofits “VHL Family Alliance has meant the world to me. Through this alliance I have met friends who can relate to the unique world of VHL. Questions, concerns, and ideas about VHL have been through this organization. Many thanks and kudos to the VHL Family Alliance. Truly, you have helped me climb the mountain of VHL. THANKS!!!!” — amy19004

“VHLFA...this is the most amazing organization who supports all whose lives have been affected by this rare condition. Questions are answered and support given freely and with care. If you have VHL or if someone you love has VHL, you will never walk alone as long as you are a part of this group.” — myway

<http://greatnonprofits.org/organizations/browse/search:VHL+family+alliance>



Special Thanks to our Hotline Volunteers!



Myriam G.

Every day, one of our Hotline volunteers stands ready to speak with you on the phone. The Hotline volunteers are on the front lines of providing support. They are available throughout the day and evening to be a friendly listening ear, to support you in times of stress, and to answer your questions about VHL. Questions range broadly—from what VHL is, to where to find medical help, to queries about the latest information on nutrition or clinical trials.

These are wonderful people who have learned a lot from their personal experiences and are glad to share with you what they have learned. They are good listeners, and if they don't know the answer themselves, they will help you get the best answer.

They are your entry point into a chain of information that stretches literally all around the world. It consists of the chapter leaders, the country leaders, the Clinical Care Centers, and our Medical Advisory Board, all ready to help you.

When you call you are likely to speak with one of these wonderful people:

Jim C. from Maine—our newest hotline volunteer, has been dealing with VHL for 30 years, diagnosed when he was in the military. He is now retired from the military and from a subsequent career in fire and police. He now lives where you have to drive 25 miles to find a stoplight!

Jule F. of Texas—Jule and her daughter have VHL. Having lost her mother when she was 6, Jule knows what it's like having sad VHL memories, but is generating hope and modeling good health management for her child. She dreams of a cure for her child.

More Quotes from Great Nonprofits

"I was diagnosed with VHL 4 years ago, and if it wasn't for the VHL Family Alliance, I don't know what I would have done. I'm from a country where the disease is extremely rare, the information is almost nonexistent and my life would have been a constant battle if I wouldn't have benefited from the help of VHL Alliance Family and its members. Great job and keep up the good work! Thank you!" — monaxa

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Jim G. of Washington State, a proud veteran of the U.S. Military in addition to being a VHL veteran—Jim works in telecommunications and spares time to talk with you. He has recently had a successful brain surgery in Seattle.

Myriam G. of Maryland, who answers in Spanish—Myriam has relatives with VHL in Spain. She has been a volunteer with us for more than 15 years, has helped to translate many of our materials into Spanish and has worked with the growing network in Spain.

Jody H. in South Carolina, who has VHL herself—Jody has a delightful sense of humor and a cheering outlook on life. She has participated in a clinical trial.

Robin K. of Texas—Her husband and two sons have VHL. Robin teaches elementary school and is the primary coordinator of doctor appointments and strategies for her family.

Kathy M. in New Mexico, a teacher of English as a second language and a vigorous supporter of her students as well as others in the VHL community—Kathy and her brother have VHL.

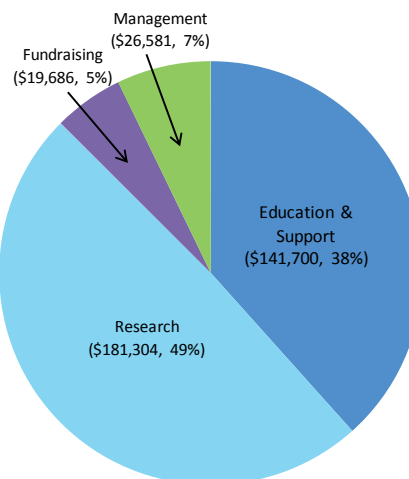


Robin K.

Fred T. of New Hampshire whose daughter Stephanie has VHL—Fred has been active in creating and running support groups in New Hampshire, and in working with Dartmouth Hitchcock and other medical centers to raise their awareness of VHL. A former Telephone Pioneer, he and his motorcycle buddies raise money for VHL.

Thanks to our Hotline volunteers, conveyers of HELP and HOPE!

Report for Fiscal Year 2012 (ended June 30, 2012)



Total Expenditures for FY2012: \$369,271
Total Revenue for FY2012: \$385,233
Net Assets: \$175,068

Thank you to MD Anderson for supporting the 10th International VHL Medical Symposium with an in kind gift of \$127,915!

VHL Research: Leading to a Cure

by James Gnarr, Ph.D., member of the VHLFA Board of Directors and Chair of the Research Advisory Board



As we learn more about how VHL operates within the cell, we are ever closer to finding a cure. Among other roles, VHL is a key player in the process of Angiogenesis. Angiogenesis is a normal process involving the growth of new blood vessels from existing blood vessels, but when a tumor develops it is often because the cancer cells communicate with the blood vessels to stimulate abnormal angiogenesis.

Before the VHL gene was identified and well before we started to have an understanding of the function of the VHL protein, it was suspected that the loss of function of VHL in tumors would play a role in angiogenesis, because of the vascular components of VHL tumors. We now understand that a key function of the VHL protein is to suppress the expression of another protein called hypoxia-inducible factor, or HIF.

HIF is a “master regulator” that controls the expression of a large number of genes whose protein products stimulate many activities in the cell, including the stimulation of angiogenesis. These findings led to the development of many clinical trials over the last several years involving newly developed inhibitors of angiogenesis, which initially focused on patients with non-hereditary renal cancers.

Today, trials using angiogenesis inhibitors are being tested in a variety of cancers and VHL patients are now

being included. Intensive laboratory research efforts are ongoing, searching for new and more effective angiogenesis inhibitors, as well as direct inhibitors of HIF activity.

Another area of VHL research that has received much recent interest involves the epigenetics of cancer cells. While the genetics of a normal or cancer cell refers to the DNA sequence of the genes that are contained within that cell, the epigenetics of a cell refers to which genes will be expressed in a cell. The human genome is made up of more than 3 billion (3,000,000,000) base pairs on 23 chromosomes that range in length from about 8.5 centimeters (chromosome 1) to about 2 centimeters (Y chromosome). If the chromosomes were laid end-to-end, they would span more than 2 meters. This DNA has to be packaged neatly into a cell nucleus that is about 6 micrometers (0.000006 meters) in diameter. The textbook analogy is to take a fine string of 40,000 meters (24 miles) in length and insert it into a tennis ball.

The epigenetics of a cell refers to the organization of the DNA that is packaged in the cell nucleus, which is mediated by several families of proteins. These proteins respond to signals to help determine which genes will be active by making them available to the gene expression machinery or inactive by keeping them inactive. Recently, mutations in the genes that encode some of the proteins that are involved in packaging the DNA have been identified in renal cancer patients, and research to determine how these mutations affect the choices of genes that may be expressed or silenced in cancer cells is ongoing.

Clinical Trials: The Hope for a Treatment

by Eric Jonasch, M.D., member of the VHLFA Medical Advisory Board and Chair of the Task Force on Clinical Trials

There are several clinical trials open or about to open for people with VHL. Most of these studies are using agents that were developed to target the tumor blood vessels (endothelium) and inhibit angiogenesis.

Continued development of these agents means that side effects are going down and effectiveness is going up. We are hopeful that people living with VHL benefit from this crop of “next generation” drugs in the near future. It is pretty clear that different people respond differently to each drug, and one of the things we are trying to learn is how to predict who will benefit more from one drug than from another.

A recently reported study showed that VHL patients treated with Sutent (sunitinib, Pfizer) showed a consistent decrease in the size of kidney cancers. Based on that study, a new trial has opened using a similar drug, Votrient (pazopanib, GSK). This study is currently open at MD Anderson Cancer Center in Houston, Texas, and will open soon at University of North Carolina, Chapel

Hill. Patients are eligible if they have genetically proven VHL, or have a clear clinical picture of VHL, with multiple sites of organ involvement, and areas that can be measured on either MRIs or CT scans. Patients receive Votrient for six months, and have the option to continue at that point if it is considered appropriate for the patient. Scans are performed prior to starting Votrient, after three months of therapy, and after six months of therapy.

A second study is testing dovitinib (Novartis) for people with VHL who have hemangioblastomas. This study is opening in October 2012 at MD Anderson Cancer Center in Houston. Patients can have other VHL related lesions, but must also have at least one hemangioblastoma. This study is based on research suggesting that one of the proteins that is blocked by dovitinib is a driver of blood vessel growth in hemangioblastomas. Treatment will be for six months, and scans will be performed at the start of treatment, after three months, and after six months of treatment. If you have questions or would like to join a trial, please call Cherie Perez at 713-563-7252.



Please help us cure VHL!

We Need You and Your Help Funding our Urgent Efforts...

Thanks to a challenge grant from the Lusk, Greene, and Horwitz families, your donation from now until the end of the year will be met dollar-for-dollar up to \$30,000!

In order to continue to Care, Share, Learn, Research, Educate and help to Diagnosis VHL, we need your involvement.

Here are some of the immediate ways **you** can make a difference:

DONATIONS: All donations received through the end of 2012 will be matched. Your contribution directly supports VHL patients and medical research. Support us with your tax deductible contribution today.

FUNDRAISING EVENTS: You can organize an event to raise funds for VHLFA. A patient's sister, Stephanie Kaes, organizes a softball tournament in Canada every year to raise funds. This year, they raised \$25,000! See

a list of ideas at <http://www.vhl.org/ways-to-help/fundraising-ideas>.

ATTEND VHLFA EVENTS: See our calendar <http://www.vhl.org/patients-caregivers/get-involved/calendar-of-events-and-meetings/>.

CREATE A LEGACY: Consider making a bequest of cash or property to VHLFA in your will. Call our office to obtain our charitable non-profit information.

COMBINED FEDERAL CAMPAIGN: All federal employees may designate the *Alliance for Cancer Research and Support – VHL* as the recipient of their generous gift by indicating our five-digit charity number 10934. Many states have state-level campaigns as well. See the brochure for our number.

Thank you in advance for your generous contributions to VHLFA; we can't do it without YOU!

Mail to: VHLFA, 2001 Beacon St, Ste 208, Boston, MA 02135-7787 USA
or Canadian VHLFA, 4227 Hamilton Rd., Dorchester, ON N0L 1G3

Enclosed is my tax-deductible gift to support: ☐ VHLFA ☐ Research only
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(Please make checks payable to VHL Family Alliance)

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