



**Annual Report issue,  
2010-2011**

*Family, Friends, Physicians, & Researchers dedicated to improving diagnosis, treatment, and quality of life for people affected by von Hippel-Lindau.*

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# The Time is Right for Advancement in VHL Support and Research

by Jeanne McCoy, Chairman of the Board

It's been just under 20 years since scientific researchers identified the VHL gene. This landmark discovery has led to ongoing explorations on how the genetic flaw which occurs in people with von Hippel Lindau results in tumor formation. Since the discovery of the gene, 3300 scientific articles were written on VHL, 6% of which were published within the first 9 months of 2011! Furthermore, along with this increased interest in VHL, scientific methods, diagnostic tools, and medical treatment for many types of cancer have also advanced.

Yet we, as members of the VHL Family Alliance, know there is much more work to be done! More doctors need to be educated about VHL in order to properly diagnose the disease and provide medical care. More research is essential in order to develop and offer therapies that are even more effective to VHL and non-hereditary kidney cancer patients. Past research grants provided by the Alliance (\$1,300,000!) have contributed to the progress we have seen in recent years. Nevertheless, we are aware that medical research is expensive and that the Alliance needs to increase future funding. We need your financial support more than ever. Over the next five to ten years we anticipate major breakthroughs in these areas, bringing to light increased awareness, better diagnostic tools, much more effective treatments, and, yes, hopefully even a cure!

It is up to us, the consumers of VHL knowledge, to heighten the

momentum. Moreover, until the day arrives that a cure is found, we need to be there for each other. We need to support each other during times of critical need and to be there for those who are recently diagnosed and searching for answers.

According to our mission, *the VHL Family Alliance is dedicated to improving diagnosis, treatment, and quality of life for individuals and families affected by von Hippel-Lindau disease.* As Chairman of the Board of the VHL Family Alliance, I encourage you to help us, your "other family", to help you. How can you use your time, talent, and treasures to promote the Mission? Together we can maximize the momentum occurring in the medical community as we continue and expand our efforts in....

**SUPPORT:** We have an international network of family support groups. We can get help in person, on the telephone hotline, over the internet, and in personal gatherings.

**EDUCATION:** We learn from each other and with our physicians, medical teams, researchers, and general public including the VHL Family Alliance Annual Meeting and VHL Medical/Research Symposium sponsored by the Alliance.

**RESEARCH:** We provide financial support to key medical research towards developing a cure for VHL and by providing tissue samples to the tissue bank.

**DIAGNOSIS AND TREATMENT:** We work with and encourage VHL Clinical Care Centers throughout the world.

**WE NEED YOUR HELP!**

***"We want to make sure that you get the best possible treatment today and to ensure that better treatment options are available as soon as possible."***



[bbb.org/charity](http://bbb.org/charity)



## Surviving VHL – A Poem— Staples part II

by Keith “Just Sayin” Richards from his book, *Animated Poems, Volume I*

Here we go again. This time it’s eight years later.  
This time it’s in more than one critical place so the stakes are even greater.

I had checkups before so I knew what was to come wasn’t attractive.  
But this time I knew what I was up against so I was proactive.  
Doctors here in New York weren’t working fast enough so things were already looking kind of bleak.  
But thanks to my mom, I was up in the hospital by the end of the week.

I got myself situated for yet another milestone in my life.  
And early Monday morning, there I was once again under the knife.  
Coming out I had headaches like you wouldn’t believe and my strength was on a decline.  
But compared to any other outcome, this will suit me just fine.  
Mom and Dad were there every single step of the way to help me through the pain.

To relay messages and prayers and just to keep me sane.  
My sister kept me encouraged to hurry up and get better as time is taxing.  
And that I have too much work to do to just sit around relaxing.

Like I said, my strength was dwindling but as time passes I get back more and more.  
As I recover I can say that every day is better than the day before.

*In cannot thank my friends enough for coming to see me and keep me strong.  
They kept me smiling and laughing so the days didn’t seem so long*



*I received phone calls every day from friends and family with such nice things to say.  
Sincerity was overflowing as they just checked to see if I was okay.*

*Everybody’s adopted me as their own so naturally they all want the right things to be done.  
It’s funny because this type of thing brings out the doctor in everyone.*

*“You don’t want to do anything to get yourself stressed.”  
“Don’t you dare rush back to work.” “Make sure you get plenty of rest.”*

*It’s pretty amazing how a turnaround was made so soon.  
For me to go through major brain surgery on Monday morning and be homebound by Saturday afternoon.*

*People have asked me what I’d like for my birthday this year.  
As cliché as it sounds I’m just happy to be here.*

*I was never afraid of the outcome. My whole attitude was “come what may.”  
Because the very God that I served then is the same one that I serve today.*

*I’m fully aware of whose I am and I’ll never lose sight of who sits on the throne.*

*It’s a very humbling experience to know that the life your life is so much bigger than your own.*

To buy the book, visit: <http://www.buybooksontheweb.com/px?kw=animated%20poems&typ=Title>

To visit Keith’s website, visit <http://www.justsayinmusic.com/>

## Remember VHLFA in Your Will

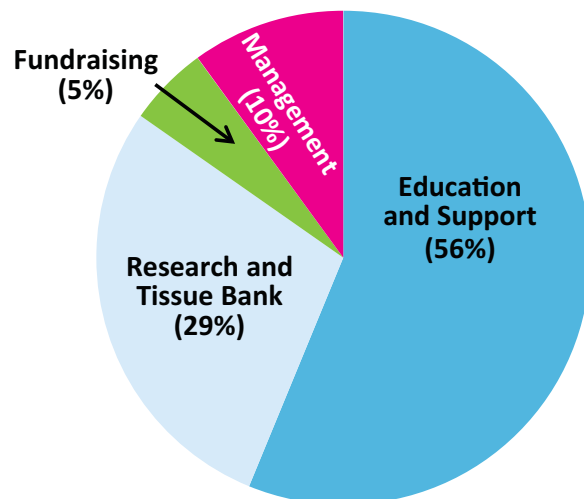
You can give hope to millions of people worldwide with VHL, kidney cancer, and other tumors by extending your support of VHL Family Alliance programs beyond your lifetime. Whether your legacy is large or small, you can support our research and educational programs by remembering VHLFA in your will.

To make a bequest of cash or other property to VHLFA, please set up a meeting with your attorney and provide him or her with the following information:

VHL Family Alliance, Inc., a non-profit corporation organized under the laws of Massachusetts  
2001 Beacon Street, Ste 208, Boston, MA 02135  
Federal tax ID 04-3180414

A bequest to VHLFA is fully deductible for estate tax purposes. In addition, remembering VHLFA in your will is an important and personal way of providing hope to people with von Hippel-Lindau disease for generations to come. You may wish to learn about other gift opportunities by consulting with advisors, or simply send e-mail to [director@vhl.org](mailto:director@vhl.org), or phone Ilene Sussman at 617-277-5667 ext. 4.

## Report for Fiscal Year 2011 (ended June 30, 2011)



Total Revenue for FY2011 was \$253,653

Total Expenditures for FY2011 was \$246,629

**Please help us cure VHL!**

# Research Projects Your Dollars are Supporting

## Disease-Causing Mutations

A grant was awarded for Eleanor Rattenbury, a second-year Ph.D. student in the laboratory of Dr.



Eamonn R. Maher, University of Birmingham, England, to study the DNA changes that alter the function of the VHL protein and inhibit its ability to suppress cell growth.

Not all VHL gene DNA changes will cause VHL disease and so if a patient in whom VHL disease is a possibility, but not certain, is found to have a DNA change that has not been detected previously, it is often unclear whether that person has VHL disease (in which case their relatives are at risk and they may develop further tumors) or if it is just a rare but benign DNA variant. In such circumstances it would be very helpful to have a test (or tests) available that would reliably establish whether the change is disease-causing or not.

This project seeks to develop tests that will directly assess the functions of the VHL protein and enable predictions regarding whether the DNA change causes disease or not, and if it is a disease-causing mutation, which tumors are likely to develop. This will not only be of practical benefit for patients undergoing VHL gene testing but will also inform on the relationships between the function of the VHL protein and tumor development.

Better understanding of this relationship will be helpful for developing new therapies for VHL disease.



## Understanding Drug Resistance in Renal Cell Carcinoma

A grant was awarded for Dr. Haifeng Yang of the Cleveland Clinic to study Drug Resistance in clear cell renal cell carcinoma (ccRCC). Mutations of the *VHL* tumor suppressor gene cause von Hippel-Lindau disease as well as sporadic ccRCC, the most common kidney cancer in the general population.

Historically ccRCC has been resistant to all forms of traditional chemotherapy. VEGFR inhibitors generated positive clinical outcomes but not cures, so more drugs against new targets are needed. Suppression of a potent oncogene EGFR significantly blocked kidney tumor growth in mouse models, but EGFR inhibitors failed in kidney cancer patients.

Recently it was found that a histone-modifying enzyme JARID1C was mutated in ccRCC. They found that when there is no VHL protein in the cell, JARID1C was responsible for the global reduction of an important epigenetic marker and suppression of some HIF-responsive genes. While HIF2 $\alpha$  is an oncogene in ccRCC, promoting tumor growth, JARID1C is a tumor suppressor. When JARID1C was knocked down in ccRCC cells in a mouse model, the tumor grew significantly. HIF-dependent JARID1C activation also led to innate drug resistance to EGFR inhibitors in VHL-deficient kidney cancer cells.

Dr. Yang will investigate further the role of JARID1C and explore ways to defeat resistance so EGFR inhibitors could become useful drugs to VHL patients.



## Give to VHLFA while Shopping on the Internet

### *iGive, a Simple Way to Give!*



iGive.com is a service that provides a percentage of your online purchases to the charity of your choice. Their new and improved "button" is a convenient way to make

these donations without changing your searching and buying behavior at all – no special sites to access, you don't have to think about it at all.

You can use your favorite browser, no toolbar is added. The "button" software watches to see what site you are going to, and indicates the sites that

participate in iGive.com. Make your purchase as usual, and iGive will send us a check!

Please give it a try. It costs you nothing, and will make an important and painless donation to help us find a cure for VHL!

Go to <http://www.igive.com/> button to install this utility. One installation puts it on all your browsers, on Windows or Mac. Register us as your favorite charity by searching for "VHL" and choosing VHL Family Alliance to make sure your donation comes to VHLFA. **THANK YOU!**



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

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 2011 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. Ten year-old Alex Anderson raised \$4,000 with his lemonade stand! See a list of ideas at [http://www.vhl.org/aboutvhlfa/fundraiser\\_ideas.pdf](http://www.vhl.org/aboutvhlfa/fundraiser_ideas.pdf).

**ATTEND VHLFA EVENTS:** See our calendar <http://www.vhl.org/meetings/index.php>.

**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 VHL Family Alliance 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 magnanimous contributions to VHLFA, we can't do it without YOU!

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**Mail to: VHLFA, 2001 Beacon St, Ste 208, Boston, MA 02135-7787 USA**  
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Enclosed is my tax-deductible gift to support:  VHLFA  Research only  
 \$25  \$50  \$100  \$150  \$500  \$1000  \$ \_\_\_\_\_  
 My employer will match my donation. I have enclosed the necessary forms.  
*(Please make checks payable to VHL Family Alliance)*

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- Send newsletter
- Audio version needed
- I would like to receive occasional alerts via e-mail from VHLFA only

I am a  Person with VHL  Family member  Friend  Sponsor  Health professional \_\_\_\_\_ (specialty)

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Name as it appears on the card \_\_\_\_\_ Exp Date \_\_\_\_\_

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*VHLFA is a non-profit corporation in the U.S. and a registered Charity in Canada*

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