



## **Educating and Empowering People with VHL**

I just returned from the 9<sup>th</sup> International Medical Symposium on von Hippel-Lindau, this year held in Rio de Janeiro, Brazil, hosted by the Brazilian National Cancer Institute (INCA) and chaired by Dr. Jose Claudio Casali da Rocha, head of the Brazilian National Tumor Bank, and co-chaired by Dr. Eric Jonasch, Genito-Urinary Oncologist, M.D. Anderson Cancer Center in Houston, Texas. We have held a medical symposium every two years beginning 1994, moving it around the world in order to attract participants from various countries. This year's meeting was the first ever held in Latin America.

A complete report of this meeting will soon be available on the internet and in the January newsletter. Meanwhile, let me share a few of the highlights to demonstrate the importance of our efforts to educate and empower patients, families, and physicians in the United States and worldwide.

There were ten of us from VHL families in the U.S. and Europe meeting with 40 patients and family members from Brazil and Argentina. All of us were struck by the differences in our experiences. Where the representatives from Europe were healthy people with a few surgeries under their belts, the people from Latin America had been devastated by VHL. It is only in the last 5-10 years that they have a diagnosis, that they have any idea what is happening in their bodies, or how to protect themselves.

For many, this meeting was their first introduction to how genes work. Some families felt they were "cursed," that they "must have done something bad" to have so many medical problems. Their doctors did not know what to do. Even once they were told it was VHL, their doctors could find no more than "two paragraphs in 1400 pages of information about rare diseases."

***You can help inform doctors in the U.S. and worldwide.***

That's where our website comes in. That's why our Handbook is so very important. Available free for download from our website in fourteen different languages, it is the first and most important document for helping families and their local doctors find problems early, and seek the best treatment at the right time.

***We need all our friends to help us fight VHL. Education is the first step.***

Elena told us the story of her eldest son who has had a series of brain tumors. No one knew why there was more than one tumor, they thought their family was cursed. And then he had a spinal tumor, also with no known cause. Then a young geneticist from São Paulo suggested a DNA test for VHL. At last there was a name for the condition, but still the doctors didn't know what to do. Her other two children were tested for VHL, and both were positive. Elena was terrified. But as she met others with VHL, read the Handbook, and sat through the lectures at the Symposium, she began to realize that she and her doctors now have tools to protect her other children. The family now understands that VHL is not a curse, it's a genetic alteration, and that everyone on the planet has several genetic alterations.

***You can help people learn their DNA status and how to protect themselves!***

Karen in the U.S. was diagnosed with VHL and has been managing her health well for years. Her brother began to have symptoms that Karen was sure were also VHL. His doctors discounted her theory and refused to do an MRI, feeling it was not warranted. His symptoms persisted, and Karen was increasingly certain it was VHL. They begged for an MRI, but were denied. Finally he passed out and was taken to the emergency room, where they finally did an MRI - and found multiple tumors in his brain. Now it was certain it was VHL. VHLFA worked with this family to empower them to persist in negotiations with the medical system and get to the source of the problem. "The VHL Family



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



Alliance helped save my brother's life last year. Thank you! Thank you! Thank you!" -- K.M.

***You can empower families!***

We distributed copies of the Kids' Handbook in Spanish to all attendees. This book was designed for the many children known to have the DNA alteration but who in most cases have not yet had a VHL medical issue. It helps parents and children have a positive conversation about VHL. The Spanish translation was done by Dr. Karina Villar from Barcelona, Spain. Carlos Fredes, the volunteer chairman of our affiliate group in Argentina, has agreed to distribute materials throughout Spanish-speaking Latin America. Volunteers in our affiliate countries have already produced translations of the Kids' Handbook in Spanish, German, and French, with Dutch and Portuguese nearing completion. This gentle introduction to VHL is causing a great deal of excitement. It is short, colorful, and full of hope.

***You can help us enroll children early into preventive screening and keep them healthy!***

We distribute information through the Handbook, the Newsletter, the recorded lectures on the website, and through online support communities in English, French, German, Spanish, and Japanese.

The Handbook and Screening Protocol outline for general practitioners how to monitor the health of the patient and find any problems early, when they are likely to be more successfully treated, and to give the family time to assemble the best team to treat the presenting issue.

***You can help print and distribute Handbooks!***

On Wednesday before the symposium began a group of twelve physicians and consumer advocates from seven countries met to review the Handbook, and especially the screening protocol, to update it with the learning of all countries to make it more effective, more economical, and easier for patients to accomplish in their busy lives. The suggested revisions will be presented in the January newsletter, and the revised Handbook will be published in 2011.

***You can help continue to improve the clinical guidelines through collaboration!***

The first day of the Symposium was devoted to Basic Science, reporting advances by teams across the globe in understanding the series of events that occur in the cell when there is too little VHL protein to suppress tumor growth. Among these events the researchers are identifying a number of new "molecular targets," or places where a drug might be used to intervene, get the cell processes back in order, and stop or shrink the tumor.

***You can help continue to fund research projects by young scientists, introducing them to the fascinating study of VHL.***

The Symposium was a celebration of the teamwork we have created together – physicians, researchers, and families. We also need the help of all our friends and supporters to help us fund this work, increasing awareness, making materials and information available in print and on the internet, in multiple languages, and continuing to strengthen and learn from the global community.

***You can help maintain the momentum, continue to improve management of VHL, and find a cure! – Thank you!***

**Holiday Gifts**



Instead of one more tie or bottle of perfume, how about a gift of research to find a cure?

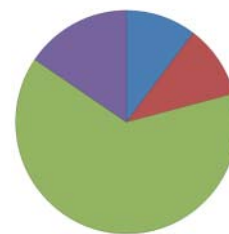
We will be happy to send you blank cards to send yourself, or we can send cards to the recipients you ask us to honor. Minimum \$10 per card.

This year's card is a picture of one of the illustrators of our wonderful new *VHL Handbook Kids' Edition*, now available in English, Spanish (European or Latin American), French, and German!

**Report for Fiscal 2010 (ended June 30)**

*preliminary numbers - audited numbers available 11/30/2010*

Total Revenue for Fiscal 2010 was \$279,177. Of this, \$40,437 was allocated to the Research program, for grants and Tissue banking.



*Of our Expenses:*  
 Research - \$40,437 (15%)  
 Education & Support- \$166,061 (63%)  
 Fundraising - \$27,728 (11%)  
 Management - \$26,769 (10%)

**Please help us cure VHL!**

## Challenge! Your Gift Goes Farther!

Four families are challenging you to make a special pledge to VHLFA this season. They will double your gift, up to a total of \$55,000! and help us raise money for research. Special thanks go to Scott & Amy Borg, Sunny Greene, Lee and Toni Horwitz, and Tom and Nancy Lusk for their generous Challenge Grants!

## Research efforts you are supporting 2009-2010

Dr. Brenda Petrella, a Research Biologist at the Veterans Administration Medical Center in White Junction, Vermont, is receiving funding this year for her project to evaluate the effectiveness of a new approach to treating kidney cancer. If it works, it would not only benefit people with VHL, it would also benefit all people who get "sporadic" kidney cancer – acquired at random in the general population. Most sporadic kidney cancer tumors have an alteration in the VHL gene that occurred during their lifetime and caused the tumor. It is the lack of sufficient VHL protein in a cell that causes a kidney tumor to start.



Her team has identified a new therapeutic target, MT1-MMP. When VHL function is lost, levels of MT1-MMP are significantly increased in kidney cancer cells. MT1-MMP regulates several cellular processes, including those leading to the development of tumors. There is evidence that kidney cancer cells use MT1-MMP to enable them to gain the capability to metastasize, or seed themselves in other parts of the body.

Their hypothesis is that inhibition of MT1-MMP

may therefore have anti-tumor effects in kidney tumors and may represent a novel therapeutic target for the treatment of kidney cancer, which is highly resistant to most chemotherapy. In this study, they are working to determine whether inhibition of MT1-MMP has anti-tumor effects and whether inhibition of MT1-MMP increases the efficacy of sorafenib (Nexavar), a currently FDA-approved first-line therapy for advanced kidney cancer that has spread to other organs.

The results from this study will contribute to our knowledge of the molecular mechanisms driving the development and spread of kidney tumors. We are hoping that this research will help us to find effective non-surgical treatments for kidney cancer tumors for people with VHL and for others in the general population who have VHL alterations in their kidney cancer tumors.

### Tissue Banking

Research requirements for tissue banking are changing. Much research these days requires that samples have their RNA intact. RNA degrades rapidly as soon as tissue is removed from the body. Ideally, tissues need to be flash frozen in or near the operating room. We are working to understand the requirements of the researchers, and with our Tissue Banking partner, NDRI, to understand the best methods for acquiring, transporting, and storing tissues that will meet the researchers' needs.

## Feedback from families with VHL

See these and many other comments at ...

Having a rare disease can be overwhelming! Because VHL is so unique, no one can really understand the questions and concerns you have. Finding the VHLFA was a godsend. -- G.A.

This organization has helped me and my husband more than words can say. VHLFA has provided key information that has improved his quality of life and given us tools to help his doctors continue to check and prevent future health problems. It reassures us that we're not walking this road alone. -- D.L.



<http://greatnonprofits.org>



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[www.guidestar.com](http://www.guidestar.com)



[www.cancercureamerica.org](http://www.cancercureamerica.org)  
- search "VHL"



together we're better™

[vhl.inspire.com](http://vhl.inspire.com)

## Your Donation Goes Even Farther!

Thanks to the generous support of the Borg, Greene, Horwitz and Lusk families, *Your donation* this season will go even farther!

Sunny Greene, Scott and Amy Borg, Lee and Toni Horwitz, and Tom and Nancy Lusk have challenged us. They will match all contributions that we receive this season, up to a total of \$55,000. Please help us earn these wonderful donations!

We're offering you something you can't buy at any price -- life-saving information to help people stay healthy with VHL, and targeted research projects working for a cure for VHL.

You can send holiday greetings to your friends and relations that will help match these gifts! Fill out the form below, or list addresses on other paper, to send holiday gifts *and* help us find a cure.

**Gifts of the Artists:** Several talented members of our community have donated CDs and books. For a donation of \$150 or more, you can choose one of these wonderful performances as your free gift.

**Thank you!**

## 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 [development@vhl.org](mailto:development@vhl.org), or phone Joyce Graff at 617-277-5667 ext. 4

**\*\* Write in the item number on your donation form.**

1. "VHL Handbook, Kids' Edition", by Kruger et al
2. CD, Bach, "Goldberg Variations" for Piano, by Pierre Jacomet
3. CD, "Forever In His Care," by Deb Hogan
4. CD, "Wayfaring Stranger," by Clenton Winford II
5. CD, "Swing Favorites," by Shannon Forsell



Mail to: VHLFA, 2001 Beacon St, Ste 208, Boston, MA 02135-7787 USA  
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*(Please make checks payable to VHL Family Alliance)*

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