

## Improving our Understanding of Pancreatic Tumors

At the Boston meeting in June 2007 Dr. Steven K. Libutti of the National Cancer Institute, shared with us a snapshot of the state of his 12-year study of pancreatic lesions in VHL. He has been screening the VHL patients in the NCI study to see how many in fact have pancreatic lesions, and then closely reviewing all cases with pancreatic lesions.

"If it were not for the VHL community," Dr. Libutti said, "and the advocacy of the Family Alliance, and the proactive nature of the people with VHL, we would not have been able to accomplish this study . . . You need to salute each other for the data that I am sharing with you."

As a result of this study, NCI is changing its recommendations for when and how to treat pancreatic lesions. This is new information that you will not find in the 2005 edition of the *VHL Handbook*. An article is expected to be released in the December 2007 issue of the *Journal of Surgery*, making the new recommendations official. Meanwhile, he shared what he described as "a work in progress," which may change yet again prior to publication. Her paper has been submitted to the journal *Nature*, acknowledging the support of the VHLFA.

About 75% of people with VHL develop cysts of the pancreas and about 12% develop solid tumors. The cysts do not require action at all unless they lead to compressive symptoms. Some of the "hard tumors" turn out to be microcystic adenomas, honeycombed clusters of small cysts, that look solid on the scans but in fact are not a problem.

But some people do get serious tumors, Pancreatic NeuroEndocrine Tumors (PNETs), which can progress to cancer in rare cases. These need to be taken quite seriously, but not all of them require intervention. In fact, only 8% of the PNETs they have seen have metastasized. The goal of his study has been to understand these tumors better, and to see if he could find some additional characteristics that would help to guide the medical team in choosing if, when, and how to intervene.

Size is traditionally our best measure, and remains an important indicator.

If a PNET were actively generating hormones that were causing symptoms, that would be an important factor. However, among the 108 VHL patients with solid lesions suggestive of PNETs in the NIH study he has not found a single functional PNET. If anyone out there, anywhere in the world, has a functional pancreatic tumor that is generating hormones and causing symptoms, Dr. Libutti wants to see you. Seriously.

They looked at gender to see if there was any correlation. The solid PNETs were evenly divided among men and women.

They also examined the DNA. They found a higher correlation of dangerous PNETs among people who have an alteration in Exon 3 of the VHL gene. The VHL gene has three distinct parts, called Exons. Each family has a particular mutation, like a misspelling of one word in the book of instructions that make up the VHL protein. That family mutation is passed intact from parent to child, so each family member has the same alteration in their VHL gene. People with a mutation in Exon 3 seem to have a more aggressive type of pancreatic tumor.

They also looked for signs of aggressive behavior. To measure aggressiveness, they took a series of images and compared the size of the largest tumor in each of these scans, then calculated its rate of growth, or "doubling rate". If the tumor doubled in size in less than 500 days, it was deemed to be high risk. If it took longer than 500 days for the tumor to double, it was at a more moderate risk level.

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In the past, recommendations for when to operate have been based entirely on size. But now, with the addition of these new measures, he has divided tumors into three categories – low risk tumors can be watched every 2-3 years; medium risk tumors should be followed more closely, and high-risk tumors should be evaluated for surgery. (see Figure 1)

Dr. Libutti is looking for more people with pancreatic tumors to help validate his findings. He encourages anyone with a hard tumor of the pancreas to apply to be part of the NCI study. The person to call is Geoff Seidel, his clinical research nurse, at Tel: 301-496-5049; Fax: 301-402-1788.

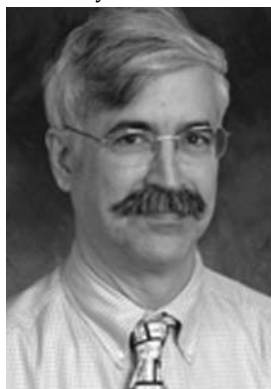
High risk - evaluate for surgery	Medium risk - follow until a second criterion is present	Low risk - follow every 2-3 years
Size $> 3$ cm	Size 2-3 cm.	Size $< 2$ cm.
Mutation in exon 3	Mutation in exon 1 or 2	Mutation in exon 1 or 2
Doubling in $< 500$ days		

Figure 1, assessing the risk level of a pancreatic neuroendocrine tumor. – preliminary findings

## Report from the Boston Meeting

The 2007 Annual Meeting was held in Boston, Massachusetts, on June 23-24, 2007. This report contains only the highlights. All presentations have been recorded. A CD or DVD is available for purchase, or you can view selected segments on the website, for a modest fee. We hope that you will benefit from these excellent presentations. See <http://vhl.impactlearning.org> or mail your order using the coupon in the back of this issue.

Two of the researchers YOU have been supporting for the past year with your research donations were on hand to share their research. **Dr. Susanne Schlisio** of Dana Farber Cancer Center described her research on the molecular causes of pheochromocytoma. She studies some important clues: first that the risk of pheo varies depending on where in the VHL gene the mutation occurs. Second, VHL is one of five different genetic alterations that can result in a pheo. In all cases, the genetic variations disrupt the normal cycle of creation and elimination of Egln3 and KIF (pronounced eggelin-3) during embryonic development. These left-over cells from before birth hang around in the body and later cause a pheo to form. Her team is working on translating this understanding into treatment.



Richard Cohan, M.D.  
Radiology  
University of Michigan,  
Ann Arbor

**Dr. Donald Bellgrau** of the University of Colorado shared his research on targeting cancer-specific mutations (like VHL) with Immunotherapy. All people develop tumors, but the immune system normally kills the tumor. The immune system looks for foreign material (like a splinter) and launches defenses against this foreign material.

It tests to see if cells are “self” or “not self”. A second analysis determines whether this material is dangerous. Cancer has the ability to elude the immune system by fooling the body into believing that this tumor is “self”, or even if it is “not self” it is determined to be “not dangerous.” Bellgrau’s research is working with a yeast preparation that can be used to “wake up” the immune system and sound the alarm that the cancer cells are indeed “not self” and are “dangerous”, spurring the body to mount a defense against the cancer cells. In cooperation with the Cleveland Clinic they have prepared a customized therapy that will work for some of the VHL mutations. This compound has to be customized for each different VHL mutation, or category of mutations. The project continues, with second year funding from VHLFA.

**Dr. Richard Cohan** of the University of Michigan spoke with us about issues in imaging for VHL. Dr. Cohan specializes in imaging of the abdomen and pelvis. For VHL, this means imaging of the kidneys, pancreas, and adrenals. He discussed the differences among conventional X-ray, ultrasound, CT, MRI, and PET. Contrast material is important in a CT: it makes tumors brighter but does not change the appearance of a cyst. There is probably no more increased risk for people allergic to shellfish or topical iodine than for people who have other allergies. He shared ideas for managing people with sensitivities to the contrast materials.

MRI is often preferred, especially in young people, because it does not use radiation. MRI gives excellent images and better contrast than CT, but the image resolution is not quite as good as CT. There is concern about the use of gadolinium contrast agent with people who have elevated creatinine levels, an indicator of poor kidney function. This is something that should be discussed with your urologist. Most people with VHL are at extremely low risk for complications with gadolinium.

## Recuperating

**Amy Lynn Budd**, Artist in Residence at the Perishable Theater in Providence, Rhode Island, shared with us her story of having a brain tumor of VHL. After surgery she went back to studying dance, and found that it helped her regain her balance and self-esteem. She now teaches dance to battered women and women who have had breast cancer or other surgery, to help them feel beautiful again. Amy got us up and doing some easy stretches and fun moves to 60's music.

## Pheochromocytoma

The next three presentations focused on diagnosis and treatment of pheochromocytoma. **Dr. Arthur Tischler** of Tufts New England Medical Center helped us understand what pheos are, and how to diagnose them. Common symptoms and signs include two or more of:

- High blood pressure (sustained or spiking)
- Headaches
- Palpitations
- Visual disturbances
- Constipation
- Anxiety, panic attacks, mood swings...
- Night sweats or heavy sweating

80% of pheos occur in or near the adrenal glands. The other 20% are paragangliomas that occur in other parts of the sympathetic nervous system. These are sometimes called extra-adrenal pheochromocytomas.

Most doctors have been taught in medical school that 10% of pheos are extra-adrenal (outside the adrenal gland), 10% are hereditary, and 10% are malignant. However, this is proving not to be true. Up to 30% of people with pheos have a hereditary risk factor. The risk of malignancy depends on which of the genetic factors may be at work. The good news is that the risk of malignancy is low in

people with VHL. Nonetheless, pheos are very damaging tumors that need to be diagnosed and removed as quickly as possible.

**Dr. Ronald Lechan**, also of New England Medical Center, elaborated on diagnosis of a pheo. To the list of possible symptoms he added:

- Pallor (flushing)
- Nausea, vomiting

Attacks can last seconds to hours, and can be as infrequent as once every few months.

Untreated, pheos can lead to devastating consequences: heart attack, stroke, fluid in the lungs, and circulatory collapse (shock). Pheochromocytoma can be associated with metabolic abnormalities: lactic acidosis, hypercalcemia (elevated calcium), glucose intolerance and diabetes, and associated syndromes (ACTH and VIP).

The most reliable test for a pheo is measurement of metanephrines and normetanephrines in blood and/or urine. Drs. Lechan and Tischler like to do both studies to see how they correlate. Each of the hereditary conditions has a particular biochemical phenotype -- a different set of "normal" characteristics. (See Figure 2).

MRI or CT of the abdomen may identify visible masses, but they do not measure the function of the tumor. For function, there are some nuclear medicine tests that can be used: MIBG, Octreoscan, or fluorodopamine PET. The definitive management of a pheochromocytoma is surgery to remove the tumor, preferably by laparoscopy. "Pre-operative pharmacological blockade of adrenergic receptors is mandatory": that is, before surgery they have to give medications to block the receptors for the chemicals produced by the pheo. These blockers may need to be given for weeks before the surgery, to ensure that there will not be a pheo crisis during the surgery.

Drs. Tischler and Lechan, and Dr. Jonathan Coleman of Memorial Sloan Kettering Cancer Center

	Multiple endocrine neoplasia type 2	von Hippel-Lindau syndrome	Sporadic
number of cases	35	56	169
N/F	16/19	31/23	86/83
Mean age +/- SD (years)	41.0 +/- 12.2	47.3 +/- 16.0	41.3 +/- 15.8
Plasma-free normetanephrine (%)	86	96	98
Plasma-free metanephrine (%)	100	11	60
Plasma-free noradrenaline (%)	41	71	82
Plasma adrenaline (%)	44	4	43
Urinary normetanephrine (%)	100	95	94
Urinary metanephrine (%)	95	14	54
Urinary noradrenaline (%)	52	78	82
Urinary adrenaline (%)	58	2	38
Urinary vanillylmandelic acid (%)	63	36	77

Figure 2: Different profile for pheos in each hereditary condition. Pacak et al., J. Int. Med. 257: 60, 2005

in New York then listened with all of us to four patients telling their stories of a very long and rocky path to diagnosis of a pheochromocytoma.

**Jim B.**'s mother had a pheo during her pregnancy (age 20, 1953) when he was born. She survived that one, and had a second pheo at age 42, and a third at age 44. When Jim was 9 in 1962 he had three pheos, one of which was behind his heart. At age 15 he had another pheo near his adrenal gland that caused a collapsed lung. Both Jim and his mother received excellent care for their pheos, but no genetic diagnosis was made. Beginning in 1992, his eldest daughter had pheos at ages 9, 15, and 17. In 2000 a second daughter had another pheo. A total now of 11 pheos over 47 year span, and no genetic diagnosis. It was not until 2005 when Jim experienced blood in the urine that he was finally diagnosed with VHL – now with advanced tumors of the kidney, pancreas, and retina. He and his daughters are now in a proactive program of screening, to find other issues early.

**Judy F.** of New Hampshire shared the story of her two sons. Her younger son was diagnosed with a pheo at age 12. It was considered sporadic, even though Judy's aunt and her daughter had both died of metastatic pheos in the 1960's. One year later her older son (age 23) began having similar symptoms. The doctor said that pheo was such a rare tumor that the odds that it would happen to the second son were astronomical, and he refused to do the pheo testing. He forgot to think about genetics. The family did find a doctor to do the test, and her son was diagnosed with metastatic pheo. Clearly something genetic is going on, but they are still unsure just what. One would have predicted that this would be a case of SDHB mutation, but both boys have tested negative for SDHB and other likely genetic causes.

**Audrey** from Canada had sent in her story of being diagnosed with a pheo after having had a stroke. Her doctor put her on blocker medications that make her spacey and keep her almost a zombie.

### ***You can hear it ALL!***

Videos of the Boston meeting are available at <http://vhl.impactlearning.org>

A CD or DVD containing all the presentations is \$50 (the same price you would have paid if you had gone to the meeting). Handouts are included on a CD in either case.

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**No internet? See page 15 to order by mail.**

She lives in fear of another stroke. The issue here is that blockers alone will not manage the effects of a pheo over time. She is working with a neurologist to determine whether she is a candidate for surgery.

**Malinda S.** from Pennsylvania told us of her 16-year saga, actively seeking medical help for symptoms that began with hypertension, sweating, and panic attacks. She was tested for many conditions, and was basically told to see a shrink. The symptoms escalated and the panic attacks became rage attacks. One doctor who worked with her later told her that he had considered a pheo, but for two reasons ruled it out: her blood pressure was high, but not spiking, and none of the descriptions talked about rage. A number of texts say that blood pressure should be spiking. Especially with hereditary pheos, it may be high all the time or in fact it can be low. And while the textbooks usually use words as strong as "angry outbursts", Malinda would use the word "rage". She lost weight and looked terrible. At 85 pounds the doctor felt that her liver seemed enlarged. The scan showed instead that it was being pushed out of place by a 9 cm pheo.

Once the tumor was removed and her body settled back to normal hormone levels, Malinda has completed a college degree and her family life is happy again. But those last three years before the diagnosis were rugged. She points out, though, that if the pheo had been diagnosed any time in those first 13 years of searching for a diagnosis, she would never have gotten to the point of rage. There were clues; there was plenty of opportunity for a diagnosis. But the available literature failed her and her doctor. There are new publications in the literature in the last 2-3 years that are trying to improve the information available to support doctors in making more timely diagnoses. The families need to help connect their doctors with this new information, some of which directly contradicts what they were taught in medical school.

On the second day of the conference **Dr. Scott Plotkin** of Massachusetts General Hospital told us about his work with Familial Tumor Syndromes, including VHL. He is working with a consortium of hospitals doing clinical trials for some of these diseases, trying to bring more options to his patients in Boston. In most cancer trials, there is one focal tumor and the objective is to shrink that one tumor. When there are multiple tumors, however, it is hard to choose one to focus on. How should we measure success? He is holding a conference in Boston November 29 on designing trials for people with tumor-suppressor syndromes like VHL, to determine what the specific goals of such a trial should be.

**Dr. Daniel Cho** of Beth Israel Hospital gave us an overview of the clinical trials available now

for kidney cancer. Most are focused on metastatic disease, which we with VHL work hard to avoid. Sorafenib looked promising, but at the end of 18 months the benefits seem to wear off. Sunitinib (Sutent) approaches the problem from more angles and is showing promise. The angiogenesis effects do seem to wear down after a year, but the effects remain, and some promising agents are coming out that may prolong the effects. Temsirolimus, for example, works against cell proliferation. All three of these agents have now been approved "for advanced kidney cancer." None has yet been explicitly approved for use with VHL. One active study is under way testing Sutent specifically for VHL. It should be noted that while there is response from these drugs with kidney tumors, there is no significant response reported for CNS tumors.

**Dr. Othon Iliopoulos**, head of the VHL Clinical Care Center at the Massachusetts General Hospital, shared his research on biomarkers for the identification of kidney cancer thru analysis of blood or urine. It would be great if there were an easy blood or urine test that could indicate the possible presence of kidney cancer – it would allow the doctor to identify people who need follow-up scans to determine whether kidney cancer were present, and it might help doctors measure the success of treatment – as the kidney tumor shrinks, the indicator levels go down; if the tumor recurs, the indicator levels go up again. A blood test is a great deal cheaper and easier than a scan.

Dr. Iliopoulos has done an extensive study of blood and urine samples from RCC patients, and has identified seven leading candidates for biomarkers – chemicals in the blood whose levels seem to correlate with levels of RCC disease. He worked to identify which chemicals rose, and which dropped, with the activity level of RCC in the body. The leading candidate seems to be CA9. Studies continue.

**Dr. Nahum Goldberg** of the Department of Radiology at Beth Israel Hospital in Boston spoke with us about Radio Frequency Ablation of VHL Renal tumors. This is an outpatient procedure performed with an image-guided probe – a very attractive alternative to open surgery for some situations. Ideally, the kidney tumors should be between 2 and 3 centimeters – large enough to be a good target, but small enough that it is still considered not to be in imminent danger of metastasis.

He explained the technology, showing how the heat probe is used, and the challenges of getting it to the right place without injuring other structures. Some tumors are harder to reach than others, and depending what other delicate structures they are sitting against (blood vessels, urinary ducts, bowel,

or muscle) it may be necessary to inject saline to provide an insulating space to protect these structures, and to keep the flow of blood or urine from cooling the heat treatment before the tumor is fully cooked.

Dr. Goldberg presented his own experience, and data shared by Dr. Brad



*Nahum Goldberg, M.D.,  
Radiology, Beth Israel Hospital,  
Boston*

Wood at the U.S. National Cancer Institute, to give us an overview of the pros and cons of using Radio Frequency Ablation to treat kidney tumors. The promise of this technology is to stunt the growth of a kidney tumor without open surgery, keeping it from advancing and metastasizing. We now have eight years of data. A total of 325 patients have been reported in the medical literature. Dr. Wood and his colleagues have treated 112 tumors in 63 patients who have been followed for an average of three years after the treatment.

They conclude that RFA and other thermal therapies have much to offer for the treatment of VHL tumors. RFA for small tumors is safe and effective and can save kidney function as long as care is taken to minimize damage to surrounding structures.

Dr. Steven Libutti, head of the VHL pancreas study at the U.S. National Institutes of Health, gave us a status update of his research. See separate report. (Page 1)

At the Annual Meeting four new members of the Board of Directors were officially elected, and awards were presented to key volunteers: to Altheada Johnson, on completion of twelve years of service on the Board of Directors; to Tom Lusk on completion of six years of service on the Board of Directors; to Alexandra Morais of our Spanish language hotline, as our Volunteer of the Year; and to Fran Mott for her continuing service as chairman for Michigan.

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### ***Free Sample!***

To give you the taste of it, you can watch Joyce Graff's presentation online for free.

We will be doing more of this kind of event.  
What would you particularly like to see?

Videos?

Powerpoint slides with audio tracks?

Audio only?

## Radio Show for VHL?

What if there were a radio show devoted to VHL  
and other rare diseases?

What would YOU like to tell the general public?

What would YOU like to hear about yourself?

***Joyce has been asked to put together a one-hour a week radio show on webtalkradio***

## What do you think?

***Send your questions and ideas to  
[power@vhl.org](mailto:power@vhl.org)***

## ***Please join us at the Houston meeting Saturday, November 17, 2007***

Update on the Sutent Trial  
Advances in treatment of Eye  
Advances in diagnosis of pheo  
Meet our Ambassadors: Caleb and Chloe  
Meet Joyce Graff

What else would you like to hear?  
Write to [us-tx@vhl.org](mailto:us-tx@vhl.org)

Our hosts:  
Dr. Eric Jonasch, Chairman  
VHL Clinical Care Center  
M.D. Anderson, Houston

Clenton Winford, VHLFA Chair, Texas  
Michelle Black, Vice-Chair, Texas

## Meetings for YOU

Please tell us how we can make a meeting that works best for YOU! Please fill out the **survey on page 15** and help us design a meeting for you.

***Please Join us in Birmingham,  
Sandusky, or Houston!  
See page 12***



*Joyce Graff traveled to Japan in June 2007 to speak at the Japanese National Society for Hereditary Tumors at the invitation of Dr. Taro Shuin of Kochi University. Joyce was one of only two presenters from the West at this all-Japanese meeting. She also spoke to a class of genetic counseling students at Ochanomizu University in Tokyo. Their professor, Cheiko Tamura, who studied at Johns Hopkins in Baltimore, was the first genetic counselor in Japan, where she is working to establish this new profession. Here Joyce is shown with three of the students, Maki, Yuumi, and Shino, who helped to host her visit.*

*Guess who's a Boston Red Sox fan?*

## VHL in South Africa

My name is Markus Jansen van Vuuren and I have Von Hippel-Lindau disease (VHL). As with many cases of VHL, my diagnosis was delayed. Although I grew up with the knowledge that my father had three brain operations to remove tumors and also laser treatment of his eyes, I did not relate my symptoms to what had happened to my father. This is probably because he had passed away of cancer of the kidneys when I was 20 years old and my symptoms only started when I was 36 years old.

My symptoms began with headaches which started as a slight pain in the forehead that shifted to the back of the head and became very intense -- feeling like my skull wanted to break open and explode! Over a period of nine months the intensity of the headaches increased, I felt dizzy, started squinting, experienced loss of balance, and -- very scary -- I also could not use my right arm. During this time the General Practitioner (GP) prescribed various types of medication for sinusitis, migraine, other pain killers, antibiotics, etc, but with no relief. Still I did not connect what was happening to me with VHL.

On the 3rd of August 2000, I went to another GP and in desperation asked for a referral to have a CAT scan done. After examining me and giving me another prescription for sinusitis (!), I was referred for the scan. The CAT scan revealed a tumor the size of a golf ball and a large cyst on my brainstem. I was referred to a neurosurgeon who requested an MRI scan which revealed another tumor of about 1cm x 0.5 cm in position T5, touching both the bottom of T4 and the top of T6 of the spinal cord. The brain



*Dr. Lizette van Rensburg, Medical Sponsor, VHL-SA*



*Markus Jansen van Vuuren, Chairman, VHL-SA*

tumor and cyst were removed on the 9th of August 2000 and the tumor in the spinal cord on the 2nd of October 2000. It was a frightening time for me and my family. I am happy that the operations were successful and that I am recuperated.

Later that year, DNA testing by Professor Lizette van Rensburg (Human Genetics, University of Pretoria) detected an altered VHL gene in my blood DNA. This information allowed testing of other members of my family to determine whether they carry the faulty VHL gene. I am currently following a screening program of annual imaging of my kidneys and a twice-yearly eye checkup, as well as MRI scans when needed, for early detection of tumors so that treatment can be carried out in a timely manner.

With the help of Dr. Van Rensburg we have published an article in a journal that is sent to all physicians in South Africa, to alert them to the signs of VHL, and most importantly to announce the formation of a VHL support group in South Africa. We are rather excited about this and hope that through the VHL-SA support group we can learn from each other, educate ourselves, the medical community and the general public about VHL. We will be affiliated with the VHL Family Alliance International, an organisation that has established an international network of family support groups.

The first meeting of VHL-South Africa will be held March 15, 2008, in Johannesburg. For additional information about the meeting, watch [vhl.org/meetings](http://vhl.org/meetings). To find out more about VHL-SA and to assist with the establishment of our own support group, please contact us: Dr. Lizette van Rensburg or Markus Jansen van Vuuren at the following telephone numbers: +27(0)12- 319-2636 and +27(0)82-779-5019 respectively or email us :

## Remember the Tissue Bank!

Please take the time to register with the Tissue Bank. Do it now – donate blood, urine, and saliva, and fill out the paperwork “just because”.

These seemingly small donations will help researchers who are looking for biomarkers – simple tests that a doctor could do in the office, like the dip-stick test for sugar in the urine. In order to find a simple test, they have to evaluate thousands of samples from people in various stages of kidney involvement – from zero to high involvement. With your help, we can improve tools for diagnosis, and for tracking the effectiveness of treatments.

In the paper issues sent to U.S. and Canada you will find a reminder magnet. Please keep this handy magnet where you will see it – on your refrigerator or in some other prominent spot. When surgery is planned, there are so many things to organize and think about – please remember to take a moment to call or write the bank. If you have pre-registered, all it takes is one phone call.

Even if you have not pre-registered, the staff at the bank will work with you to organize things quickly and efficiently.

Tissue donations are the most important elements in finding a cure. Without tissue to study, research cannot move forward.

If you have a magnet from before, you will notice that we have given you a new number and e-mail address. The old ones will work, but these are more direct and easier to remember. The new magnet has the e-mail “bank@vhl.org”

Thank you! For giving *the gift that only you can give* – and getting us one step closer to a cure!

The NDRI Tissue Bank can accommodate donations from U.S. and Canada and some other countries. You are always welcome to inquire. For countries with their own tissue banks, please contact your local bank first. See <http://www.vhl.org/bank> for details. Magnets are being sent only to members in the U.S. and Canada.

## New Clinical Care Center in New York City *Memorial Sloan Kettering*

Memorial Sloan Kettering Cancer Center has recently worked with the VHL Family Alliance and established a VHL clinical care center. Memorial Sloan Kettering (MSK) has seen and managed patients with VHL for years and the establishment of this clinical care center will serve all VHL patients more efficiently. Memorial Sloan Kettering’s clinical care center includes a coordinated team of physicians that are experienced in the comprehensive care of VHL patients. This will allow for thorough patient evaluation and enhanced continuity of care for individuals with VHL and other hereditary kidney cancer syndromes.

Jonathan Coleman MD and Cassandra Mills RN will be the patient’s first points of contact. Dr. Coleman is a urologic surgeon and worked at the U.S. National Institutes of Health (NIH) with significant clinical and research expertise in VHL including minimally invasive procedures and organ conserving surgery. Ms. Mills has a broad experience in multiple medical fields and has served as a nurse coordinator skilled in managing complex medical care. They are very interested in VHL as a whole and have organized an outstanding team of physicians and researchers at Memorial Sloan Kettering for the benefit of their patients.

Though Dr. Coleman’s area is Oncology with an interest in VHL kidney and adrenal tumors, the structure of the Memorial Sloan Kettering program is thorough in its scope. As described, “the initiative includes the creation of a multidisciplinary team to evaluate each patient for the multiple manifestations of this disease process and to coordinate the care and follow-up among the different disciplines. A shared database of clinical information will be created that will not only aid in tracking patient care but also allow for the identification of successful treatment strategies to promote continual improvements in clinical care.”

This database may prove to be one of the most useful tools to physicians who follow VHL patients at MSK. The implementation and effective use of this clinical database will serve to address all areas of concern for the patient, accessible by all members of the team. This approach will allow for early detection and tracking of medical conditions associated with the syndrome.

To contact Memorial Sloan Kettering Cancer Center, Telephone +1(646)-422-4432 or send a Fax +1(212)-988-0768. Dr. Coleman will be able to review your case and help coordinate your visit according to your medical needs.



## Join the Conversation! vhl.clinicahealth.com

*The online support conversation at <http://vhl.clinicahealth.com> is strong and wonderful. You will find great support there, a forum to ask questions about whatever is bothering you, and a warm community of people who understand what you are going through.*

*Here is one example of a posting in the online group.*

### Dig's Diary

#### First Entry

I have not been diagnosed with VHL.

I'm a man who has had cysts and tumors of various sorts my entire life. In my twenties doctors told me I was just "one of those unlucky folks" who tend to develop these things. At 37 I was diagnosed with clear cell renal cell carcinoma, which was successfully treated with radical nephrectomy.

Several months ago I had an MRI for possible multiple sclerosis due to visual disturbances I have been having ... pain with eye movement, temporary loss of vision, dizziness. No MS lesions were seen, but they did find a large cyst of some sort on the left side posterior of my cerebellum. The neurologist said that it is likely just an arachnoid cyst that many people get and is not causing my symptoms. I'd like to believe him, but the headaches and pressure are getting much worse and I have unbearably loud tinnitus in that ear.

#### Second Entry:

Last week I had an abdominal CT for my annual cancer follow-up and another nodule was found in my left adrenal gland. My doc says this is likely just a benign adenoma but has ordered an MRI for a clearer picture. He has not mentioned anything about blood work. Incidentally, I have always had low blood pressure on account of a genetic heart defect ... 90/60 is not uncommon. However for some inexplicable reason my blood pressure of late has been skyrocketing and my blood sugar is up as well when it has always been very good.

## Our Ambassadors



Chloe and Caleb E. from Texas, whose mother is a regular participant in the online discussion, have consented to be our ambassadors in this year's Combined Federal Campaign. You will see their sweet faces in our campaign ads on the internet. Chloe, age 13, is battling tumors on the optic nerve in both eyes, and her third pheo. Caleb, age 9, has had five pheos removed and now has a sixth. They are asking everyone to join us in finding a cure for VHL. If you work for the federal government, please encourage your fellow workers to give to the

**Cancer Research Fund - VHL,  
CFC # 10934**

We are also in most state campaigns. Check the brochure. If we are not in the brochure, you cannot write us in. Please send a check.

**Thank you!**

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This is all very frustrating to me. I can no longer believe I am just this "unlucky" guy who tends to develop random cysts and tumors. Although my doctors would have me believe that these are all just unrelated coincidences, my thoughts are that there is an underlying cause for this all. This is what led me here to this board.

I hate like heck to self-diagnose, but I think it's time to take the bull by the horns and look after my own health care. My next stop is to a retinal specialist to get a good look at my eyes, and I'm also going to ask my primary care doctor to order blood work to check this "nodule". Any guidance as to what specifically I should be asking my docs for would be very much appreciated.

Thanks,  
Dig62

Excerpts from postings by one member of the VHL online discussion at ClinicaHealth. What would YOU like to say to Dig? Sign up and join the conversation at <http://vhl.clinicahealth.com>

# Cure Steven! Cure VHL! Cure kidney cancer! Eat pancakes!

*from Beverly A., Florida*

Family and friends banded together to hold a Pancake Breakfast at Beef O'Brady's Restaurant in Florida to benefit VHL Research. In particular, we wanted to show our love and support for Steven, my 18-year-old son, who was diagnosed at the age of sixteen, and has undergone two brain surgeries in the past two years.

Beef O'Brady's is a Sports Bar, usually open only in the evenings. They opened on a weekend morning just for this event. I've done lots of fundraisers for worthy causes, and this was the easiest fundraiser one ever! The restaurant ran the kitchen, and the volunteers served pancakes, sausages, and drinks. We advertised in a local paper and rounded up all our friends. I sent lots of e-mails -- "come and be silly and eat pancakes with us. If you've ever played baseball with one of our children, come eat pancakes with us."

We had a balloon guy, some guessing games, and a tattoo table (washable tattoos!) to add fun and spice. We tried out VHLFA's new Frisbees. Dale and I made T-shirts for the servers to wear. The local newspaper interviewed Steven.

Altogether we raised more than \$4400 for VHLFA!

And at the end of the day, though the numbers were wonderful, it wasn't the numbers that mattered the most . . .

## ***Inspiring***

The best part of this day was having Steven take back a bit of the control that this disease tries to rob from him. The best part of the day was seeing all of the sweet people who are helping him do this.

## ***Touching***

Bottom line, Steven knows that he is doing all that he can to find a cure.

## ***Empowering***

Bottom line, he knows that he is very loved.

## ***Amazing!***

(And the numbers were pretty good too! And, of course, we're very, very thankful for all of the



*Jackson and David modeling their balloon hats*

donations and money raised through pancakes!)

## ***What if***

VHL were as well known as other rare diseases?

Just a thought, as I sit here at 2 a.m., reading through a local paper and noticing that a group of women, just a social group, managed to receive a huge donation from a bank for their fundraising project. Maybe people are more willing to donate big if they have had personal experience with the disease. Maybe it is easier to receive these sweet givings when the donor has actually heard about the disease that you're fighting for.

Wouldn't it be sweet to receive a donation so huge that it would cover supplies for five pancake breakfasts?

Just a thought.

## ***What if***

people actually realized that this fundraiser is not only for Steven and VHL, but also for most kidney cancer fighters?



*Steven, David, Mary, and friend*



*Beverly and Dale made shirts for the servers.*

Just a thought, really, as I sit here at 2 a.m., realizing that most people don't know of the relationship between VHL and the cases of kidney cancer that are found in the general population.

Renal cell carcinoma is known as a "silent killer" in the world of cancer. It has so few and such vague symptoms, that by the time it is discovered, the cancer has often metastasized to various parts of the body. That coupled with the fact that there is not a reliable drug protocol to fight this form a cancer makes kidney cancer the seventh leading cause of cancer death.

The good and interesting news is that most cases of kidney cancer in the general population are caused by isolated, sporadic mutations of the VHL gene, the sort of mutations that tend to happen as we age or are exposed to environmental cancer risks. So...whatever cures VHL will also cure these random cases of kidney cancer. That is reason enough for VHL to come out of hiding and receive the sort of public recognition and financial assistance that other causes benefit from.

Just a thought.

### ***What if***

each one of us did one thing each year to raise awareness of VHL and raise money for research?

***I'm glad to hear these fundraisers were successful. I'm proud of us who have VHL and I'm glad that this Alliance exists. Keep up the good work! Together, we will make a difference.***  
***-- Jennifer Kingston, Australia***

## ***Birmingham meeting***

***Saturday, October 13, 2007***

***See [www.vhl.org/meetings](http://www.vhl.org/meetings) for details  
or call 1-800-767-4845  
or Alabama Chapter Chair  
Liza Parker, 256 829 2305  
[us-al@vhl.org](mailto:us-al@vhl.org)***

***Bring your questions!***

***Meet new people with VHL!***

***Meet Joyce Graff!***

***Meet key contacts at the  
University of Alabama,  
Birmingham  
see also page 12***



*Steven playing the soccer he loves*

**Free Teleconference meetings:**

**Understanding NCI:** Toll-free Teleconference Series. You are invited to participate in the teleconference series to learn more about NCI's important cancer research programs and how advocates are involved. Callers will have the opportunity to ask questions.

**Wednesday, October 31, 2007, 1-2 pm EST**

Update for the Advocacy Community from the NCI Director, Dr. John E. Niederhuber and Mr. Doug Ulman, Chair, NCI Director's Consumer Liaison Group

USA Toll-free: 1-800-857-6584, passcode PCP

Toll-free playback: 1-866-443-8027 until 11/30

For more information: <http://pcp.cancer.gov>

**Thursday, November 8, 2007, 1-2 EST**

From Bench to Bedside: Research at NCI-Frederick, Maryland

1-800-857-6584, passcode: Frederick

Toll-free playback: 1-866-379-4229 until 12/9/07

**Tuesday, December 11, 2007, 1-2 pm EST**

Cancer Survivorship Research: A Dialogue with Advocates, Survivors, and Caregivers

1-800-857-6584, passcode OCS

Toll-free playback: 1-800-253-1052 until 1/11/08

For more info: <http://dccps.nci.nih.gov/ocs>

**Local and regional meetings:****13 October 2007 - Saturday****Birmingham, Alabama, Support Meeting**

see [vhl.org/meetings](http://vhl.org/meetings) or call

1-800-767-4845 for details

see page 11

**Survey  
page 15**

**20 October 2007 - Saturday****Sandusky, Ohio, Support Meeting****Sandusky, Ohio, Black Tie Gala**

support meeting in the afternoon

fundraiser beginning 6 pm for VHL and MS  
October 20, 2007, 6 pm

see below

**15 November 2007 - Saturday 9-4****Houston, Texas, Regional Meeting**

see page 6

**28 June 2007****Orlando, Florida, VHLFA Annual Meeting**

stay tuned for details!

**4-6 September 2008****Copenhagen, Denmark**

Biennial Medical Symposium on VHL

watch <http://www.vhl.org/conf2008>

## Ohio Support Meeting and Gala Fundraiser - Sandusky

VHL Support meeting and Black Tie Gala - Sandusky, Ohio, Saturday, October 20, 2007

The Ohio chapter of the VHL Family Alliance and the Black Tie Dance Studio invite you to join them for some very special events for VHL on Saturday, October 20.

In the afternoon, a meeting of the Ohio chapter will be held at the Lyman Harbor Conference Center.

Speakers will include:

Dr. Rick Visci, Obstetrics and gynecology, talking about pregnancy and VHL

Dr. Arun Singh, Ophthalmology, speaking about VHL in the eye

Katie Hoffman, Genetic counselor, Cleveland Clinic, concerning DNA testing

Joyce Graff, VHLFA

**... and wait ... there's more! ...**

In the evening, a grand evening of food & fun: the Annual Black Tie Gala, a Charitable Benefit for VHL & Multiple Sclerosis Research at Cleveland Clinic.

Tickets \$30 per person for a great evening!

**Table for 8 only \$300**

**Where:** Compass Point Ballroom of the Lyman Harbor Conference Center, 1615 First St., Sandusky, Ohio (Compass Point Ballroom) 419-62-lyman (419-625-9646)

**When:** October 20, 2007. Doors open @6:00 pm

**Corporate Sponsorships Available**

To register for the support meeting and/or the Gala, please contact [cari@vhl.org](mailto:cari@vhl.org),

**Music****Dance performance****by the****Black Tie Dance Studio****Silent Auction****Hors d'Oeuvres****Cash Bar**

## **Our thanks for donations from:**

***The thank-you list appears only in the print version of the newsletter.***

***Many thanks to all our donors, and congratulations to those who have been honored by their friends.***

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VHL Family Forum, Newsletter of the VHL Family Alliance and the Cancer Research Fund / VHL

Volume 15, Number 3, September 2007, ISSN 1066-4130

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# Meeting Planning Questionnaire

You are a valued member of the VHLFA, and your input is important. This short questionnaire will aid in planning future meetings of the VHLFA membership. You may also take this survey online at [vhl.org](http://vhl.org)

1. Did you attend the June 2007 annual membership meeting in Boston? Yes / No

If yes, please skip to question 3

2. Why did you NOT attend the annual membership meeting? (check all that apply)

☐ Topics were not of sufficient interest

☐ Had made previous plans

☐ Travel too difficult or expensive

☐ Never received meeting notification

☐ Other reason(s): \_\_\_\_\_

3. Have you attended previous annual membership meetings? Yes / No

4. What might encourage your attending an annual meeting? (check all that apply)

☐ Presentations on a certain topic (please specify) \_\_\_\_\_

☐ Meeting held closer to my home

☐ Meeting at a different time of year

☐ Other \_\_\_\_\_

5. Are you active in a state chapter of VHLFA? Yes / No / ☐ There is no state chapter in my state

6. Have you attended a state / regional meeting in the past? Yes / No

7. Would you consider attending a future state / regional meeting? Yes / No

8. Are there any topics of special interest that would encourage you to attend?

Please specify \_\_\_\_\_

9. Additional comments/suggestions related to VHLFA and its operations: \_\_\_\_\_

Your name and address (optional):

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Home phone: \_\_\_\_\_ Work phone: \_\_\_\_\_

E-mail: \_\_\_\_\_

***Please email / fax / mail your comments to us as soon as possible. We will post results of this questionnaire in an upcoming newsletter.***

***Thanks for helping, and for being part of the VHLFA team!***

## ***COUPON - Ordering a CD or DVD of the Boston meeting***

Please send me a ☐ CD (plays in your computer) or ☐ DVD (plays in any DVD device)

Please enclose \$50. Individual sessions may be ordered for \$10 each (see the report on page 2)

and be sure to fill out your shipping information above.

or set up an online learning account at <http://vhl.impactlearning.org>

## Getting married?

Now you can support the VHL Family Alliance by registering with the ***I Do Foundation***.

From honeymoons to invitations to wedding gifts to charitable wedding favors, the I Do foundation allows couples and their guests to make wedding-related purchases that generate donations for charity.

The I Do Foundation's Charity Registry service also makes it easy for guests to make donations in lieu of gifts. All of these services are available free of cost at [www.IDoFoundation.org](http://www.IDoFoundation.org).

Check it out today, and be sure to select Von Hippel Lindau (VHL) Family Alliance as the beneficiary of your charitable wedding.

## Back to School?

***Get BookSox Stretchable Book Covers and help Cure VHL!***

BookSox are colorful, whimsical stretchable book covers that make your books look great and stay clean!

The company will donate \$1 to VHLFA for every book cover we sell.

Order on their website at

<http://www.booksox.com>

or advertise BookSox in your area

You can get colorful posters to put up around your community. Call BookSox at 1-800-930-2241 or write to admin at [booksox.com](mailto:booksox.com). Give them our organization number "02445-100", the number of flyers you need, and the address to mail them directly to you.

***For lots more ideas for how you can help in your own community,  
see <http://www.vhl.org/help>  
Thank you!***

## Meetings Just for YOU!

***Please help us design a meeting that fits your needs.***

***Fill out the survey on page 15 and let us know what you think!***



Newsletter of the VHL Family Alliance

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