

Treating VHL Brain and Spinal Tumors

Reporting on a presentation by Ashok Asthagiri, M.D., for the VHL Family Alliance [See Note 1]

We have had a rise recently in inquiries about the use of Cyberknife on hemangioblastomas of the brain, spinal cord, and brainstem. These machines are being installed at a number of hospitals around the country, which are advertising their services on billboards, radio, and television, as being miraculous for all kinds of tumors.

Cyberknife is one brand of machine that delivers doses of stereotactic radiation (SRS). While most SRS machines are limited to tumors inside the bony skull, the Cyberknife is able to track its target tumor as it moves with each breath. In theory, it can be used on any tumor.

With any new treatment, however, there is a need to evaluate not only the feasibility, we need to evaluate the efficacy of the treatment. Radio Frequency Ablation (RFA) is still considered experimental to some extent because the 10-year data has only now begun to emerge. Similarly, we need 10-year data to determine whether treatment of hemangioblastoma is effective. Sure, we *can* do it, but does it do the job?

Dr. Ashok Asthagiri of the U.S. National Institute for Neurological Disorders and Stroke (NINDS), one of the National Institutes of Health (NIH), presented a review of twelve papers over the past 16 years, and the results of a study of 20 people with VHL who underwent SRS treatment of 44 hemangioblastomas, with an average follow-up period of 8.5 years. For the first time, we have a significant number of patients over a length of time that begins to show the long-term effectiveness of this treatment, compared to the standard treatment, open surgery.

Dr. Asthagiri's talk goes through the data in detail. If you are considering radiosurgery, we encourage you to listen to the entire presentation. We have summarized the key points in Figure 1. While open surgery is obviously more difficult immediately, the recovery is better defined and the tumor is much better controlled over time.

Guidelines for SRS: (Lunsford et al.)

- Tumor should be smaller than 3.2 cc

- Tumor should receive 15 Gray (15 Gy) at the edges
- Tumor should not have a cyst.

Guidelines for open surgery (Oldfield et al):

- Tumor should be mildly symptomatic. Don't wait until something doesn't work.

From the NINDS Natural History study (19 patients with 138 tumors over 12 years) we learned:

- 58 of the 138 tumors (42%) became symptomatic
- 26 of these 58 tumors (45%) were not present on the first MRI
- 6% of tumors followed a linear growth pattern
- 94% of tumors followed a "stuttering" growth pattern (see Figure 2)
- 85% of the symptomatic tumors had an associated cyst or syrinx
- 80 of the 138 tumors (58%) never became symptomatic at all

Figure 1: Summary of Effectiveness

	Open surgery	SRS
Comfort of the procedure	Uncomfortable	Comfortable
Complications first 3 mo	4-8%	1%
Complications mo. 9-12	4-8%	20%
Control of tumor at 3 yrs	92%	85%
Control of tumor at 5 yrs	92%	82%
Control of tumor at 8.5 yrs	92%	50%

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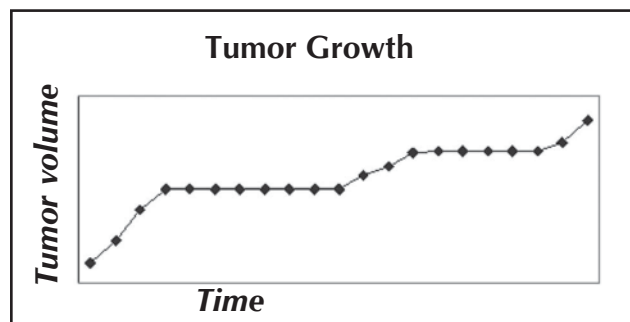


Figure 2: Stuttering Growth pattern. 134 out of 138 tumors (94%) displayed a stuttering growth pattern.

Key points to remember:

1. If someone has 10 tumors on the scan, statistically 6 of those tumors will probably never grow.
2. If someone has 10 tumors on the scan, and we treat 10 tumors, half the time we would miss the tumor that would eventually become a problem because it was not even present on the first scan.
3. If we treat a small tumor with SRS and it doesn't grow, how do we know whether it was controlled by SRS or whether it is in a quiet period of stuttering growth? (See Figure 3)
4. In the Natural History study, if they had treated every tumor that showed growth (rather than waiting for symptoms), each patient would have had an average of four additional surgeries over a 10-year period. By waiting for symptoms they were able to reduce the wear-and-tear on the patient.
5. Tumors treated with SRS (following the guidelines above) which subsequently required surgical removal showed incomplete ablation of the tumor. Some cells in the tumor had died, others had continued to grow.

Dr. Asthagiri's major concerns with SRS are:

- Near-term swelling (usually 3-18 months after treatment) may increase symptoms. Be sure to review the Questions to Ask Your Doctor before the treatment, and make a plan with your doctors for how to handle swelling if it occurs.
- Long-term lack of efficacy. There is a 50% chance that this same tumor will become a problem to you in 7 years.
- Follow-up surgery. Having to do an operation in a place that has previously had radiation treatment can sometimes make surgery more difficult.

Conclusions:

As long as the tumor is reasonably approachable, open surgery is still the first choice for long-term control. Deeper tumors, or ones on the front side of the spinal cord, are associated with more problems in surgery. Likewise, certain locations in the brain stem might be associated with more problems in surgery. You need to speak with a surgeon who

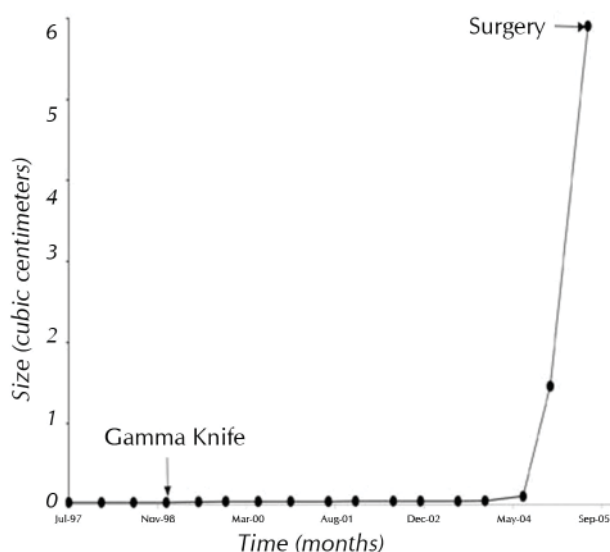


Figure 3: Recurrence. One tumor studied at NIH showed no growth for six years following radiosurgery, then grew rapidly within the next six months, requiring open surgery. Was this tumor controlled by SRS for six years? or would it have grown at all during that time even if left untreated?

has significant experience with hemangioblastomas before deciding that a tumor is truly inoperable.

It is hard to overcome the desire not to have surgery, especially if you have had multiple surgeries before. SRS is an alternative to be explored, but don't just look at the procedure itself. We also need to consider the long-term effects: it's a 50:50 chance that the tumor we treat today will become a problem to you in seven years.

The best way to improve the outcome:

- Monitor all tumors at least once every 1-2 years
- If any CNS involvement is seen, assemble your team and develop a relationship with them.
- Find the best neurosurgeon in your area, share the VHL Handbook. If needed, send scans to one of the CCC's or expert centers for a second opinion, and share that input with your local surgeon.
- Identify symptoms at early stages. Ask the doctor what symptoms to watch for, and don't be shy to call when symptoms occur. Don't quietly wait two months for the next available appointment. Make sure the surgeon is aware of the level of symptoms, and the rate of change. Tumors grow slowly; cysts can grow dramatically.

To see the video of Dr. Asthagiri's presentation, his slides, and a detailed list of references, please go to <http://vhl.org/videos/webinars/radiosurg1.php>

For the "Questions to Ask Your Doctor" when considering stereotactic radiosurgery, please see the VHL Handbook, or <http://vhl.org/stereo>

Other machines used to perform SRS: Gamma Knife, Proton Beam, Linear Accelerator, Collimator, Cyclotron, Photon Beam.

Investment in Research

In fall 2009 President Barack Obama announced a new \$5 billion investment in medical and scientific research, medical supplies, and upgrading laboratory capacity, which is expected to create tens of thousands of new jobs.

The funds will pay for “cutting-edge medical research in every state across America,” the White House said. One billion dollars will go to research into the genetic causes of cancer and potential targeted treatments. The awards will take the form of grants, meaning that institutions and researchers will have to apply for them. More than \$1 billion will go to genomic research.

More than \$1 billion of the grant funding is dedicated to research applying the technology produced by the Human Genome Project which culminated in the sequencing of the first human genome. Since then, researchers have sequenced approximately a dozen additional genomes. Now, through Recovery Act funded research, scientists will sequence over 2,300 complete genomes – or 168,000,000,000,000 DNA base pairs. This effort will allow researchers to make quantum leaps forward in studying the genomic changes linked to cancer, heart, lung, and blood disease and autism – potentially leading to new treatments and cures.

Within two years, we expect to have a chip that will permit sequencing of a person’s entire genome for under \$1000. This will allow scientists to study all the influences in a person’s body that might influence the progress of disease, or complicate treatment.

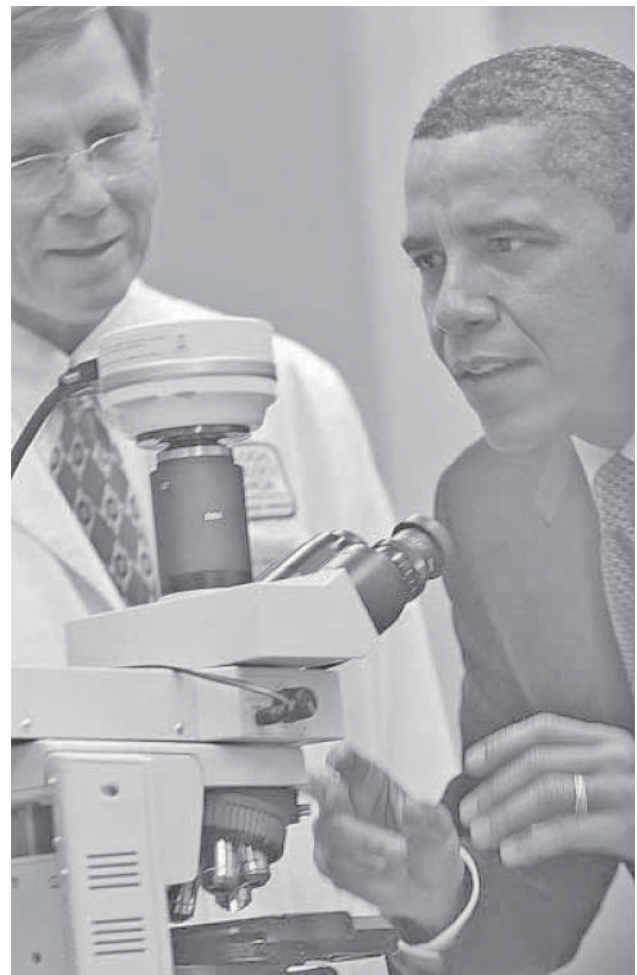
The VHL Family Alliance will also be investing in Research, both in the U.S. and in Europe, expanding and improving our tissue banking effort and our research database. Our German affiliate, under Gerard Alsmeier, will be collecting research information through a new online system, co-funded by the Robert Bosch Foundation in Stuttgart.



Gerard Alsmeier

Gerhard has led our German affiliate for more than ten years. He also serves as a consumer advocate for rare diseases in the European Union.

If the German project is successful, we hope to implement this same online system in the United States later in 2010.



W. Marston Linehan, Chief of Urologic Oncology at the U.S. National Institutes of Health, discusses cancer research with President Barack Obama. Photo whitehouse.gov

Dr. Linehan and colleagues at the National Cancer Institute have been studying von Hippel-Lindau disease since the 1980’s as part of their quest to cure kidney cancer. Dr. Berton Zbar, now retired, Dr. Linehan’s team, and the laboratory of Dr. Eamonn Maher in England identified the VHL gene in 1993.

The VHL DNA reimbursement program continues! Contact Ranjana at 800-767-4845 x4, or bank@vhl.org



Dr. Rachel Giles (right) met with more than 40 patients and family members in Begen op Zoom, the Netherlands, in October. Photo by Evert Ruis, Chairman of our Dutch affiliate

Events which touch us deeply make us strong

by Hartmut P.H. Neumann and Charis Eng, Freiburg, Germany and Cleveland, Ohio

An event in January has deeply touched our most admired founder, manager, the brain, heart and soul, and the editor of the VHL Family Alliance, Joyce Wilcox Graff: the death of her father who died at 91 years old in Memphis Tennessee January 22, 2010. We all know and love Joyce Graff, and she will be in our thoughts these days, especially when the memorial service occurs on February 20 in Memphis, and her greater family unites for a final farewell.

Professor Harry Wilcox was first a zoologist, and then became a neuroanatomist, who was an admired and beloved personality in his university. To pay their last and loving respects, his faculty and his former students, his academic family, will join his

family in remembering him.

Professor Wilcox was an inestimable help when Joyce herself was confronted at age 19, just married, with her husband's diagnosis of VHL, and for all the years later including the time when their only son was diagnosed with VHL. His knowledge and engagement was fundamental in building the VHL Family Alliance. Joyce has created this deeply desired platform worldwide. It was a key event when Joyce visited me in Freiburg in 1993, already having an idea what had to be done to understand VHL and optimize treatment using the available technology. We are all grateful to have been touched by Professor Wilcox.

Dispute over body weight presents issue to take sides on

Assignment: Memphis, by Lydel Sims

Editor's note: Lydel Sims wrote an amusing column in the Memphis paper for many years, and once featured this interview with my Dad, a good example of his wit and wisdom.

"It's amazing," the man told me, "what a group of retired men will argue about."

I don't know about that. Some people might think amazing means foolish or frivolous -- even scandalous. But what this fellow and his friends had been arguing about wasn't any of those things. Deep is more like it.

"We were discussing," he explained, "which side of the human body weighs the most."

Before you snort, try answering the question.

Which side, in fact, does weigh more? The right, or the left?

And how do you know the answer isn't significant? Did you take quarks seriously before the scientists started poking around amongst them?

All right, then.

Eager to get to the bottom (the side?) of the matter, I called the office of Dr. E. William Rosenberg, acting dean of the College of Medicine at the University of Tennessee Center for the Health Sciences.

He was at a meeting, but I left the message. Later, his office called back.

Dean Rosenberg wasn't sure, but he thought perhaps the right side weighed more in the case of a right-handed person, and vice versa, of course, for southpaws.

He hadn't commented on ambidexterity. But then I hadn't asked.

And he suggested that I consult Dr. Harry Wilcox,

who, as they say at medical college, is in anatomy.

"I don't know if anybody ever split anybody down the middle to find out," Dr. Wilcox confessed when I posed the question. I said yes, but did he have an opinion? Would the right side of a right-hander weigh more?

At this point the professor introduced a new element into the equation: compensation.

The upper half of a right-hander might well weigh more, Dr. Wilcox conceded, but one must consider the lower area of the body as well. Would there not be a compensatory shift of the center of gravity down there to keep the individual from listing rightward? Would that perhaps not result in additional weight in the lower left portion, thereby balancing the scales?

Would he say, then, that each side weighed the same?

Not necessarily. One side or the other probably weighs just a little bit more, for one reason or another, but it is doubtful that there is any identifiable consistency in the fluctuations.

That left us both pondering.

Then, I asked, didn't the continuing mystery arouse his curiosity? Would he be prepared to perform experiments to find an answer?

"Sure," he said cheerfully. "If there are any volunteers."

It was a statement of fact, but it almost sounded like a question. Nay, even an invitation. I thanked him somewhat nervously and said goodbye. It may well be that, important as they are, some questions are best left for speculation and debate.

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Meet me in St. Louie, Louie
Meet me at the ... VHL Conference!

June 19, Saturday
8:30 am - 4 pm
Marriott Union Station Hotel

What's in a Name?

by Lisa Steindel, Director of Development

The official, corporate name of our organization, as it appears on our 501C3 application from 1993, is: VHL Family Alliance. This name has served us well and does a good job describing our purpose and our mission: an alliance of family, friends, physicians and researchers dedicated to improving diagnosis, treatment and quality of life for people affected by von Hippel-Lindau Disease.

More recently, when we were approved as a participant in the Combined Federal Campaign (CFC) a national solicitation of all federal employees, it was recommended that we alter our name a bit, for purposes of the CFC campaign. As we are all quite aware, VHL is not a well known disorder and our name is not a household word. The people at CFC thought that a title like "Cancer Research Fund - VHL Alliance" would say more to the general public about what we do, and help potential contributors to find us in a list of hundreds of worthy charities. As a result, we have been the beneficiary of many federal employees who have been able to direct their charity dollars to our organization.

Unfortunately, the dual names have caused some confusion in our own ranks. Please rest assured, the "VHLFA" and the "Cancer Research Fund- VHL Alliance" are one and the same. Any checks written to one will wind up in a "single pot" with any checks written to the other. We are only one organization, funding the same research and education programs we always have.

Which name do we prefer you attach to your annual donation? It really doesn't matter. Either works for us. Our single bank account accepts contributions written to either. Much in the same way that checks written to me end up in the bank account I share with my husband. We keep it all in the family. And we know that, with your continued support, we will make our VHL family healthier and stronger - no matter which name is on the check.

"Cancer Research Fund VHL Alliance" is a registered dba name ("doing business as") with the IRS and many state registrations. You can look us up at Guidestar.org or the Better Business Bureau, bbb.org

St. Louis

Invitation from Kate Paige, Co-Chair Missouri

Please join us this June in St. Louis! We can't wait to meet you!

St. Louis is centrally located in the country and easily accessible via plane, train or car. Visit the Arch! See a Cardinals Game! Take a stroll through Forest Park (which is larger than Central Park in New York!)- Take a riverboat tour! St. Louis also offers museums with free admission, a world renowned Zoo, and a free tour at the World Famous Anheuser-Busch Brewery (see the fabulous Clydesdale horses LIVE)!!

Join us in St. Louis June 19, the Gateway to the West. Come for the conference, and stay for the fun!!!

Agenda info, see p.16 or <http://vhl.org/meetings>
 Register on the web, or on page 15.

Another Way to Help

If you own a business and would like another way to help, we have an opportunity to help each other. Glacial Energy is proud to announce that they have added the VHL Family Alliance to its Glacial Cares Program. Glacial Energy is now offering discounted commercial electricity to our family of supporters with the ability to have a portion of your monthly bill for electricity donated to the VHL Family Alliance. Your monthly electric bill can now help support research, diagnosis, education and support for families dealing with Von Hippel-Lindau. Your donation through this program adds no cost to the electricity supply and Glacial Energy saves the average business 10-15% yearly on their cost of electricity. We help each other through a simple change in supplier.

Glacial Energy is a licensed supplier of electricity in 14 states including MA, ME, NH, RI, NY, NJ, IL, MD, DE, PA, OH, MI, TX and Washington DC. They are also adding California to their ability to help businesses in just a few months. Glacial Energy is a green company with the ability to supply both standard and renewable energy products through the renewable energy credit program. If you would like to know more about how you can help the VHL Family Alliance and your business, please call Jeff Hickson BDM at 508-341-0214 or e-mail at Jeff.Hickson@GlacialEnergy.com. For more information you can also go to their website at www.GlacialEnergy.com

Smitty's Cinemas have already signed up to donate a portion of their energy bills through Glacial Energy. Many thanks to all the good folks at Smitty's! Please support the Smitty's Cinema near you. See <http://www.smittyscinema.com>

The Beginning of the Story

a Reader Story for Rare Disease Day, February 28, 2010

Editor's note: *As you read this story, see if you can guess what is wrong with this child. The path to diagnosis of any rare disease is often filled with twists and turns. The challenge for all physicians is to look at the clues, listen to the patient's story, and figure out where to go. It's not easy. And it's up to us, the "consumers," to present as many clues as possible, and to keep asking when the answer doesn't feel right.*

When I was nine years old (about 1960), I began having trouble with my eyes. I was getting one eye infection after another. More often than not, my eyes were red and felt crusty in the mornings. My vision seemed to be blurry a lot of the time. My parents would take me to the family doctor who prescribed eye drops to stop the infection and ease the irritation each time.

Their theory was that it was all part of a vicious cycle. Supposedly, one eye was infected. It was irritated, so I would rub it, and then touch the other eye, spreading the infection to that eye....back and forth and back and forth. Naturally, my vision would get blurry because of the infections.

This whole scenario and the resulting diagnosis was (and is) not uncommon for children and often even adults. Let's face it - we all tend to rub our eyes when it feels like there is something in them or our vision seems momentarily blurry, even though we've heard at least a dozen times to keep our hands away from our eyes....and for just this very reason.

The problem was that this whole situation kept going on for months, even though I really was trying to keep from wiping or touching my eyes. That's not easy when you're nine! It seemed like my vision was getting worse. The doctor blamed it on a worsening infection and finally sent me to an ophthalmologist.

The ophthalmologist had me read letters on a chart and examined my eyes with a handheld ophthalmoscope. He immediately called my parents in. He said the problem was simply that I had poor vision and needed glasses. Supposedly, I could not see things well and thus would rub at my eyes, causing them to become irritated, and the cycle would start. He said I was typical of most kids my age -- I did not want to wear glasses, and so I was not telling anyone about my vision problem. Well, I knew that was not the case! Fortunately, my parents believed me.

Not knowing where to turn next, my mother asked her optometrist to check me. I remember him being a very friendly doctor, unlike that ophthalmologist, whose gruff demeanor I remember to this day. Her optometrist thought that my left eye did not have a problem. He suspected I had something called Amblyopia affecting my right eye, making the vision poor, thus resulting in my

rubbing it a lot and causing those infections I was continually spreading back and forth between my eyes.

Amblyopia is commonly known as lazy eye. It causes reduced vision. The brain, for some reason, does not fully acknowledge the images seen by the amblyopic eye. This almost always affects only one eye but may manifest with reduction of vision in both eyes. He still was not absolutely positive, and decided to try to get an appointment for me at a large teaching and research hospital for a second opinion.

From the start of my family doctor visits, to the gruff ophthalmologist, to the optometrist, and finally to the appointment at the teaching and research center, several months had passed. Meanwhile, the vision in my right eye was decreasing rapidly.

Finally, I found myself in a huge hospital's ophthalmology clinic with lots of different doctors and medical students looking into my eyes with very bright lights. But nobody could figure out why I was losing my vision in that right eye!

By the end of that long day, I remember crying with my eyes burning from so many people looking in them, all the drops being put in them, and those ungodly bright lights. There were no retinal surgeons in those days, and the equipment they had available was not comparable to what they have today.

One of the last doctors to look in my eyes that day was a visiting physician from another country. He immediately saw the problem in my right eye. There was a tumor in my retina, directly where the optic nerve enters the eye, which was obstructing my vision. He said he had actually seen the exact same thing once in a girl in his homeland. Once he pointed out what he was seeing, everyone else could easily find it. Unfortunately, that was not the only tumor. There were several in the retinas of both of my eyes.

Hospital admission was set up. Those other tumors were treated with photocoagulation (a painful forerunner to the modern lasers.) I'd leave the hospital in a day or two, always looking like I had been in a bad car accident with one side of my face all swollen and black and blue. The ophthalmologist was invited to stay in the United States and joined the hospital's teaching staff. I had many more photocoagulation treatments in the years to come as he would find new retinal tumors in my

yearly exams. He told me the tumors were called hemangiomas.

The optic nerve tumor was more problematic though! With so much time having passed trying to get appointments to begin with, the vision in my right eye was only a blur by then and actually very distracting for a young person wanting to ride a bike and play. A tumor board met and the advice was to cut the optic nerve. Not knowing much about optic nerve tumors in those days, they were concerned the tumor would spread along the optic nerve into the brain. [See note 1] The decision was made, and that is what was done. All went well.

Remarkably, my right eye looked exactly like my good left eye after all was healed. Even though there was no vision at all in the right eye, not even light or dark, the pupil moved exactly in conjunction with the left eye's pupil. This was something that amazed many ophthalmologists seeing my blind eye for many years to come. However, eventually as the years passed, my blind eye began having other troubles. Besides regularly growing more retinal hemangiomas in both eyes, there were small amounts of fresh blood continually leaking into the retina of the right eye. Soon, they could not see into the retina. A decade later, my right eye developed glaucoma. This caused severe headaches and necessitated a surgery. Years later, the eye itself became painful, resulting in another procedure, an alcohol block, which solved that problem. Over time, and from all the work done on it, the eye began to develop a cataract and also form some large engorged blood vessels, visible on the surface. It now became obvious to anyone who saw me that my eye either had some serious problem or perhaps had been injured.

During those years, I had gotten engaged to the wonderful man who was to become my partner for life. On numerous occasions, I took him with me to the ophthalmologist, who had taken such good care of me all these years, to hear first hand about my health condition... an eye tumor condition he called dual retinal hemangiomatosis. I was concerned that if we decided to have children when we married, they might grow these tumors like I did when I was young. I wanted to take all the necessary precautions to be sure they would not lose any vision, like I did. He assured us it was very unlikely that my children would develop these same tumors. However, I was insistent. Seeing that I was so worried, he agreed to examine them at birth and yearly "until they were older and no longer at risk." He was a very skilled

Note 1: Because they did not have a correct diagnosis to work from, these doctors were making their recommendation based on a guess. We now know that this is not true. Each tumor is a separate new growth, This treatment would not be recommended today.

surgeon and a kind and compassionate man who erroneously believed that these eye tumors only occurred in young people. He thought that if one or two showed up, if treated in time, there would not be the continual progression and reoccurrence of them that I had experienced. There was never any mention of von Hippel-Lindau, or of possible other tumors in other locations. We were never told this condition could be hereditary. Information on VHL was scarce in the medical textbooks and literature in those days. It seemed that everyone thought I was just being a worry-wart.

We married in 1973 and eventually had a little girl. A year later, our son was born. We had traveled a considerable distance to that same hospital for both births so that my ophthalmologist could come over and examine their eyes before they even left the hospital for home. I was told it was not necessary, but the caring ophthalmologist who had saved my vision in my left eye and watched me grow up, agreed to it. We took them yearly for retinal exams and all appeared well. At the ages of 7 and 8, he told us they no longer needed to be examined because if they had not developed any eye tumors by now, they would not. I was not comfortable with that, recalling that I had lost my vision at ten years of age, so he agreed to keep watching them.

One day, my son developed a nasty cold. I took him to his pediatrician, only to find the doctor was out sick himself and a resident was filling in for him. The resident checked out my son and was very curious about my right eye with its poor appearance and large blood vessels on the surface. I explained all I had done over the years. I told her I had "dual retinal hemangiomatosis" and had been having both of my children followed yearly by my ophthalmologist, just in case one of them ever got this. She explained that "dual retinal hemangiomatosis" was not the name of a disorder, but only a description of the kind of tumors I had been growing in my eyes. I had always asked my ophthalmologist to send a letter to each pediatrician my children had over the years, to explain the condition I had and why they were being regularly examined. She looked in the doctor's files and found my ophthalmologist's letter. It stated that I was being followed for "dual retinal hemangiomatosis" and had requested my children be watched also, even though their risk was very low.

She told me that she had recently read an article

Note 2: Melmon and Rosen article was published in 1964, increasing awareness of VHL, and good articles were available on ophthalmology. %% add a reference?

about a rare disorder called von Hippel-Lindau and it sounded very similar to my eye history. She instructed me to call my ophthalmologist and ask him if it could be called that. I was to get back with her and let her know what he said. I did as she suggested. He said "it could be," but did not seem overly concerned. When I let her know his response, she immediately arranged for me to be seen in a genetics clinic.

I met with a genetic counselor who took a thorough medical history and family history and put together a family tree with all the information I could provide. Next I was seen and examined by a geneticist who had previously looked through all my medical records. They explained their suspicions of von Hippel-Lindau Syndrome (VHL) and a little bit about it. The VHL gene had not yet been identified in those days and DNA testing was not available, so they ordered CT scans of my brain, spine and abdomen. (They did not have MRIs back then.)

I had another appointment when the results came in. They explained that I had VHL. Together they explained all about VHL and its possible implications for my family and me. We were all scheduled for retinal exams immediately. Retinal specialists were fairly new in the eye care field back then. It was in the same teaching hospital, so my ophthalmologist went with me to that first appointment. My children were each examined also, and a hemangioma needing immediate treatment was seen in one of my daughter's eyes. Because I had already been diagnosed with VHL, this finding was sufficient for a clinical diagnosis of VHL for her. She was ten years old. My son did not develop anything in his retinas until a year later, unfortunately confirming a VHL diagnosis for him also. This was the beginning of our journey with VHL.

With the guidance of the geneticist and genetics counselor, we immediately got a medical team together to care for us, who were familiar with VHL...at least as much as could be in that day and time. They made sure our monitoring was sufficient and current, and interpreted the results for us, sending us to whichever specialist we needed when issues came up. They coordinated our care and still do. They are still keeping us alive and well as we deal with those retinal hemangiomas and all the other VHL issues that typically occur in our family.

We did not have the advantage of an organization like the VHLFA to offer us support and walk us through this step-by-step in those early days. I remember the tremendous feeling of relief and hope when we first learned such an organization had started - and to find out we were not alone dealing with VHL!?! So here it is, nearly 50 years after my original eye tumor diagnosis and my children and

I have good doctors, good care coordination and good support. We have just about everything we need....except that cure! The VHLFA is the only patient support and information organization funding research to help us with that. As the late Paul Harvey would say, "...and that's the rest of the story."

Smitty's Cinemas are donating a portion of their energy bills to VHLFA through Glacial Energy. See article on page 11. Many thanks to all the good folks at Smitty's! Please support the Smitty's Cinema near you.

See <http://www.smittyscinema.com>

You are Not Alone

by Elise, Indiana, pat of a conversation at the VHL online support group at vhl.inspire.com

"I don't have VHL, but my husband does. I was a rambunctious woman before I met him 5 years ago. I first didn't know he had it, but always wondered what that scar on the back of his head was. Then he told me.

We were one of those couples who got pretty serious, fast. And then, things were put on hold when he had to have another brain surgery a few months into our relationship. I found it so weird that this young man, who loved me, despite my imperfections, was always smiling, happy go lucky and just genuinely loved everyone around him, had this "VHL." After his brain surgery, he was still smiling. Two in a half years later, we were married, two weeks after that, he had kidney surgery. Still smiling. Two years after that our daughter was born (December 08') and now, the day before her 6 month birthday, he will be having another kidney surgery. He's still smiling, but something in him has changed. He doesn't think so though.

One thing I know for sure, is that he has always had God. Always leaned on Him in times when he's alone, always prayed and whole heartedly believed. God gave us free will. And what we do with that is our own choice. We can take the good with the bad. Or we can just wallow in our own self pity.

I look at my daughter everyday and thank God for giving me my husband. Tumors and all! I truly believe that his "condition" has shaped him into the loving husband, brother, son, father and friend that he is today. He doesn't take life or anyone in it for granted.

Just remember that you're a beautiful person and have so much to offer this world. Just remember we are all imperfect.

VHL in Russia

Dear Joyce, January 7, 2010

I just wanted to thank you, VHL Family Alliance, and all the VHL patients for supporting my recent trip to St. Petersburg, Russia for the Second International Conference on Kidney Cancer. Although the trip was short, it was an unforgettable experience.

The conference lasted only two days: one day for the patients and the other one for the doctors. I had a privilege to give three lectures on Surgical Management of Renal Cell Carcinoma (RCC), Advances in RCC Treatment, and Management of the VHL patients. All lectures were very well received by both patients and physicians. There were many excellent questions, positive comments, and an unbelievable interest in the progress of RCC treatment in the United States.

When I showed a couple of short video clips of robotic renal surgery, some commented that we practice not just in another country, but on another planet. While accepting these comments as compliments, I realized how fortunate it is to practice medicine and surgery in the United States, and how much we have to give to patients and physicians from other countries.

Many other positive comments came after my short review of the new targeted therapies for RCC. I stressed the contributions of the VHL Family Alliance, as well as long-standing contributions of Dr. Linehan, Dr. Zbar, Urologic Oncologic Branch at the NCI, and, of course, all the VHL patients who participate in clinical trials. The entire world now uses medications for treatment of advanced renal cancer that were developed based on understanding of the molecular pathways regulated by the VHL: Avastin, Sutent, and more to come.

I would like to express my sincere gratitude to all the efforts of VHL Family Alliance and our VHL patients. The difference can only be made when all



A picture of me (right) with professor Demidov from Moscow (left), while we were on a panel during one of the sessions of the conference.



Left to right: me, Carolyn Konosky (Director, Kidney Cancer Association), Dr. Ilya Tsimafeyev (Kidney Cancer Research Bureau)

the efforts are combined, and the VHL community is certainly a wonderful example of these collective contributions.

Warmest regards and best wishes to all in this New Year!

Gennady Bratslavsky, M.D.

Senior Staff, Urologic Oncology Branch, National Cancer Institute, National Institutes of Health, Bethesda, Maryland, U.S.A. Dr. Bratslavsky, a native of Russia, spoke in Russian at this conference and helped us making contact with physicians who have patients with VHL in Russia.

Clinical Trials Open

There are three clinical trials open for people with VHL. In all three cases, candidates must have one measurable solid kidney tumor, of any size.

People with tumors of the brain, spinal cord, and eye may be included as long as they meet this requirement for a kidney tumor, since the primary focus of the trial is to measure the effectiveness of the drug on kidney tumors. Nonetheless, its effects on other tumors are very much of interest. If you consider your kidney tumors "not a problem" but another problem more urgent, it is still worthwhile to speak with these trial coordinators.

To inquire about these trials, please contact:

Dr. Eric Jonasch, Urologic Oncology, M.D.
Anderson, Houston, Texas. 832-633-4519 Trial of Sutent for VHL

Sally Fowler, R.N., Urologic Oncology, 301-435-6255. Trials of drugs from Astra Zeneca and Aveo for VHL.

Dr. Ram Srinivasan, urologic oncologist with the NIH team, will report to us in St. Louis on the status of these trials.

VHL Awareness May 8 Nominations are open

February 4 was declared World Cancer Day by the International Union Against Cancer. February 28 is Rare Disease Day. May 8 this year is VHL Awareness Day.

Each of us is a Survivor, and yet we know that there may still be another round, another tumor that needs attention at the right moment in order to be successfully treated. VHL is a cancer risk factor. We control it through awareness and management.

We are not alone in our concerns about health. Everyone has some risk factors at work. One person in ten is dealing with some rare disease, and another one in ten will get cancer.

You can help yourself and your friends and relations know their own cancer risks, get a medical checkup to understand them and take action now. Here is a suggested message from the Lance Armstrong Foundation which you can easily tailor for someone with VHL in particular, but everyone on the planet is at some risk for some kind of cancer. We all need to manage our health.

Dear Friend,

It's World Cancer Day and I'm joining the team at LIVESTRONG to spread the word about the global cancer crisis and the simple things we can do to protect ourselves.

So here's the message:

Know your risks. Call your doctor....What are you waiting for?

Learning about your family history and talking to a doctor about your daily habits can give you valuable insight into the steps you need to take, right now. It's true – talking to your doctor about cancer can be scary, and many of us just forget to bring it up. But your health is important to me and that's why I'm asking you to start the conversation now.

All of us can take some simple steps, like this one, to fight cancer. For help in taking that first step, DARE TO CALL YOUR DOCTOR:

Want to nudge your friends to call their doctor? Drop the soda? Stop smoking? You can do it here:

Just send a copy of this message to the people you care about and help fight cancer in your own family and community.

There is a wonderful mythical law of nature that the three things we crave most in life -- happiness, freedom, and peace of mind -- are always attained by giving them to someone else.
- Peyton Conway March (submitted by Alice C.)

The leadership of the VHL Family Alliance is made up of people with passion -- like you! -- who move the organization forward and guide its path.

We are looking for some additional leaders who will join our team and work with us to build upon this strong foundation.

Won't you join us? Please submit your name, or the name of another person you feel would be a great addition to the team.

Please write to nominations@vhl.org

Tell us about your nominee, how to contact them, and the strengths and talents they would bring to the Board.

Deadline: March 20, 2010

DNA reimbursement continues!

The DNA reimbursement program we piloted last year was so successful that the Board has decided to continue it as a regular program of the Alliance.

VHLFA will reimburse \$100 toward the cost of DNA testing in return for your willingness to register with the NDRI tissue bank and share with us a copy of your DNA report.

By doing so, you are providing genotype information for any tissue samples you donate to the bank. This makes your samples of even greater interest to researchers, and helps stimulate their interest in studying VHL.

Please contact Ranjana with any questions: write to bank@vhl.org or call 800-767-4845 ext 4.

Thank you!

Hope - It's In Our Genes!

The last day of February is worldwide Rare Disease Day.

The National Organization for Rare Diseases (NORD) and its European sister organization (EURORDIS) are heading the celebration. VHL is doing a number of awareness meetings worldwide.

<http://rarediseases.org>

A new effort, Global Genes Project's Rare Disease Awareness initiative, is launching at the end of February. Check there for activities that you might be able to leverage in your own community to promote awareness of VHL.

<http://globalgenesproject.org>

Thank you for meeting the challenge!

The thank-you list appears only in the print edition

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Annual dues: \$25 per mailing address (\$35 outside U.S.* credit card preferred for international payments) \$ _____

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☐ Go Green! Please send all materials by e-mail to reduce costs and save the earth too!

Register for the St. Louis meeting, 8:30 to 4 pm, Saturday, June 19:**

☐ One registration (includes lunch and breaks) @ \$85 if booked by May 8 (or \$95 thereafter) \$ _____

☐ Registration for two people (includes lunch and breaks) \$155 before May 8 (\$175 after) \$ _____

** Make your own Union Station Marriott reservations, 314-421-6655, group rate \$129 (regularly \$169)

☐ VHL Handbook, Kids' Edition Members, \$6.50 ea, Non-Members \$12.99 each \$ _____

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Remember VHLFA when you want to celebrate an occasion or a loved one (minimum \$10 per honoree)

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Banking fees to process checks in non-US funds are now \$35 per check.

Canadians, please send checks to:

Canadian VHL Family Alliance, c/o Jill Shields, 4227 Hamilton Road, Dorchester, Ontario, N0L 1G0 Canada



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Regional meetings: Memphis, March 6

Dr. Robert Sanford, Neurosurgeon from St. Jude's, and **Dr. Tiffany Seagroves**, VHL researcher at UT, will talk with VHL families in the Memphis Mid-South area on Saturday, February 27, at 1 pm at St. John's Episcopal Church, Central & Greer Streets, Memphis.

Boston, April 11

Dr. Tien Hsu of Boston University, previously of the Medical University of South Carolina, and a former recipient of a VHLFA grant, will update us on the status of his research

Sunday, April 11, 2010, 1 pm. The room has yet to be assigned. Please contact the office at 800-767-4845 ext 4, or check vhl.org/meetings

Argentina, May 7

The first meeting of our Argentine affiliate will be held May 7 in **Buenos Aires**, in Spanish language at the Centro Médico de Lomas de Zamora.

Details will be posted at vhl.org/meetings or write to Carlos Fredes, argentina@vhl.org

If you are planning to attend from the U.S., please advise director@vhl.org
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USA National Meeting: St. Louis, June 19

VHLFA Annual meeting, St. Louis, Missouri
Saturday, June 19, 2010

Marriott **St. Louis Union Station** Hotel
unionstationmarriott.com, 314-421-6655

Confirmed speakers include:

- Teresa Deshields, Ph.D. psycho-oncology
- M. Gilbert Grand, MD, eye issues in VHL
- Adam Kibel, MD, urology

Registration: \$85 per person, or \$155 for two, if booked by May 8. Register on page 15, and make your own hotel arrangements. Please see vhl.org/meetings for the latest information.

International Medical Symposium on VHL

Othon Palace Hotel, Copacabana Beach,
Rio de Janeiro, Brazil

Scientific meetings October 23-24, 2010

Patient meeting Sunday, October 25

The meeting will be translated for speakers of English, Spanish, and Portuguese.

Scientific abstracts may now be submitted to the conference chairman,

Dr. Jose Claudio Rocha, conf2010@vhl.org
additional details will be posted at
<http://vhl.org/conf2010>