



VHL Family Forum



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VHLFA Research Database

by Patti K., California, Chair, VHLFA Research database feasibility study

The VHL Family Alliance is building a research database to further the knowledge of researchers, physicians, and patients about von Hippel-Lindau disease.

The VHLFA is an ideal clearing house for gathering this information. We have the largest database of VHL patients' addresses in the world. We are in a unique position to collect a vast quantity of data which will attract researchers and which, most importantly, will eventually improve diagnosis, treatment, and quality of life for people with von Hippel-Lindau disease.

In June 1994 the Alliance began examining the feasibility of such a database. Last year data was gathered on an experimental basis from fifty volunteers who attended the California state meeting and the Boston VHLFA Conference, plus participants from Spain, Australia, and New Zealand. Information from more than 60 people was entered into a trial database. This helped us identify some changes we needed to make to the questionnaires.

We are pleased to include the revised questionnaire with this newsletter. Patients are invited to participate in submitting data about VHL, and physicians are encouraged to distribute the questionnaire to any patients they may have. We hope that you will take the time to fill out the questionnaire, to add your comments about the questionnaire itself, and to add any questions you would like to see in next year's revision.

Confidentiality of patient information is of utmost concern to the Board of Directors of the VHLFA. Each individual is assigned a identification code, and information in the research database is identified only by code. Name, address, and other personal details are kept entirely separate. The VHLFA volunteers who handle the questionnaires sign confidentiality agreements. Only data from patients who have given their consent will be entered into the research database.

Summary Statistics

may be made available in this newsletter, and on our home page on the internet, as a demonstration of the kind of things we can learn from the database. For a sample of summary information, see the article on page 6. Statistical information (without patient identification) may be given out to researchers for the purpose of journal articles, statistical studies, etc., at the discretion of the Research Management Committee and its advisors. See <http://neurosurgery.mgh.harvard.edu/vhl-fa/>

Access for Researchers. The VHLFA will actively encourage researchers to apply for access to the information stored in the research database. Procedures for accessing the data will appear in the newsletter and on our home page on the Internet.

Requests to contact patients directly will be reviewed by the Research Advisory Council and members of the Medical Advisory Board. If the request is approved, VHLFA staff will determine which patients fit the researcher's criteria, and will notify them of the project or provide contact information, as the patient has requested on page 7. If participating patients prefer not to be contacted, their information will be used in the statistical studies only.

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Goals of the Database. The goal of the database is to encourage further research into VHL and to encourage scientific articles to be written for journals. Susan Booker, Research Program Coordinator for the Hereditary Colorectal Cancer Registry (HCCR) at Johns Hopkins Medical Center states that the HCCR is "just a gold mine for people who want to learn about the disease." Three years ago, the majority of articles about VHL in the literature talked about 1-3 cases. A few multi-center studies now contain information about 50-100 patients. By gathering statistical information on larger pools of patients, we hope to gain greater perspective on the range of issues in VHL, the effectiveness of various treatments, and potentially the correlation between types of mutations and types of tumors in VHL.

Seeking a Home. We are still looking for a permanent home for the VHLFA database. We have filed some applications for funding, and hope to establish it in a permanent home within the year. Meanwhile, we are expanding the pilot project to demonstrate the value to the families and researchers of having this central repository of information.

Many thanks to Patti Kohlen for her essential groundwork on the feasibility and structure of the database, and to Bill Dickson for his ongoing efforts in building and maintaining the database.

Further information. For further information, or to apply for statistical extracts, please contact the VHLFA Research Management Committee, William C. Dickson, Chair, vhlfres@usa.pipeline.com, 10613 Creamcup Lane, Great Falls, VA 22066-3306, 703-759-3665, Fax: 703-759-7992. □

Resources

N-PATH, National Patient Air Transport Hotline, the nation's only "single-source" of information on the full spectrum of long-distance patient air transport options. Call them for assistance in seeking transportation for medical care. A charitable service of Mercy Medical Airlift. 1-800-296-1217.

Sex, Love, and Chronic Illness, by Lucille Carlton. Sex has always been a difficult subject to discuss. However, it is an important part of loving relationships, and close physical contact is something we require as human beings. When chronic illness interferes with sexual issues, couples are often unable to communicate with professionals -- or even with each other -- about sexual fulfillment. Written as a labor of love, this book deals sensitively and directly with many of the frustrations facing couples dealing with a chronic illness, and gives them reassurance that they are not alone. \$13.95 through Athena Rx Home Pharmacy, 800 Gateway Blvd, South San Francisco, CA 94080 1-800-5-ATHENA. □

Ask the Experts

Question: My aunt who has VHL is a heavy smoker and enjoys an occasional whiskey. Should I worry that these habits might make her VHL worse?

Answer: We don't have statistical evidence about these habits with VHL, but we do have some statistics about cancer in general. The only thing besides VHL which is known to cause kidney cancer is smoking. It is important that people with VHL stop smoking, both themselves and others in their households. Drinking should be limited to a modest amount. Women who are pregnant should not drink alcoholic beverages. -- Berton Zbar, M.D., Laboratory of Immunobiology, Natl Cancer Inst, Frederick, Md.

U.S. Department of Health defines "moderation" as no more than one drink per day for women and no more than 2 drinks per day for men. *Dietary Guidelines for Americans*, US Departments of Agriculture and Health and Human Services, (1995), p. 40.

Question: I had an interesting test a few weeks ago. The test is called a diffusion MRI. The test showed some surprising results. My other MRI done at three different institutions showed a syrinx attached to each spinal cord hemangioblastoma. With this new procedure it shows there are no syrinx, but a fluid swollen spinal cord. With this new test my neurosurgeon was completely confused, as are the radiologists. Does this mean that every hemangioblastoma or astrocytoma with MRIs showing syrinx should have a diffusion MRI?

Answer: It has been recognized for nearly a decade that hemangioblastomas in the spinal cord occasionally cause swelling in the spinal cord that is not produced by a cyst. I believe the first report of this appeared in the *Journal of Neurosurgery* in April of 1988. In that report Robert Solomon and Bennett Stein from the Department of Neurosurgery at Columbia-Presbyterian Medical Center in New York reported six patients with diffuse engorgement of the spinal cord that was unrelated to the presence of a cyst.

With the refinement of MRI scanning and the common use of intraoperative ultrasound this phenomenon has been more completely defined. It appears to be an accumulation of fluid within the spinal cord that produces edema [swelling] and expansion of the cord and can be seen with current techniques of MRI scanning. The region of swelling often appears several months or a few years before the development of a cystic cavity within the cord (syringomyelia). Whether the spinal cord is affected by syringomyelia or swelling (without cyst) can almost always be easily distinguished by current MRI techniques. Thus, the diffusion-weighted MRI technique may help researchers understand the developmental process, but it is not essential in patient management. -- Edward H. Oldfield, M.D., F.A.C.S., Chief, Surgical Neurology Branch, National Institute of Neurological Disorders and Stroke, Bethesda, Maryland.

Beating the Odds of Inherited Illness

You can't change your genes (yet), but you can take preventive action

-- by Joyce Wilcox Graff, Editor

Barney V. knew that his father and three sisters had had their first brain tumors at 23. He was convinced that he too would have a brain tumor at 23. He got poor grades during his last year of college, and worried himself into an ulcer.

While he was in the hospital, the doctor ordered a brain scan, and found that Barney's brain was fine. He had a few small angiomas in the eye which were easily treated with laser. They gave him medicine for his ulcer, set up a regular screening schedule to monitor the VHL, and sent him on his way, much relieved.

Bob T. knew that his father had died of von Hippel-Lindau disease, but he didn't want to think about it. After all, he was young and healthy and felt totally fit -- it couldn't possibly happen to him.

It was his wife who finally pressed him to go for testing in his late twenties. Even though he did not yet have symptoms, he had a brain stem tumor which was dangerously large and which threatened to cause sudden death or paralysis. The tumor was removed, with some damage to his ability to walk. Had it been found earlier, the surgery to remove the tumor would have been much simpler.

Carol S. had watched her father die of VHL in the 1970's, and held her brother's hand through several VHL brain surgeries. She lived her life in a gloom of fatalism, fearful that each day might be the day she discovered her first brain tumor. Terrified that she might have a child with VHL, and fearful of broaching the subject with a potential spouse, she avoided forming lasting relationships.

Finally, at thirty-three, she decided to have her tubes tied so she would never have children. Her brother encouraged her first to go for DNA testing, and she reluctantly agreed. DNA testing showed that Carol does not carry the VHL gene, and cannot possibly pass VHL to her offspring. It took a year of therapy and the love of a very special partner to help her see herself as a well person with a normal future. Today she is married and expecting her second child.

Terry N. knew that his grandmother had had VHL, and that his mother and one of his uncles had had brain tumors, but he felt he couldn't afford insurance, so he didn't go for tests. Besides, he was worried about his employer finding out that he was at risk for a rare disease. When he began to have lower back pain, he went to the doctor and discovered that he had a large cancerous tumor in one kidney and several smaller ones in the other, and the cancer had spread to his lung.

VHL in the kidney is very treatable in the early stages, but once it spreads to other organs it's a much

more difficult battle. Had Terry begun preventive action on his kidneys ten years before, he would not have been dealing with cancer in the lung.

What do Barney, Bob, Carol, and Terry have in common? They were each "hiding out" from VHL in their own ways. But hiding does not protect you from VHL. It actually permits VHL to rule your life in a very different, very awful way. First, Or you may be depriving yourself of the opportunity to find out that you are in good shape and can reduce or eliminate your worries. Second, if there is something there, hiding permits the situation to worsen to a point where the outcome may more likely be poor.

“

No prescription is more valuable than knowledge. -- C. Everett Koop, M.D, former Surgeon General of the United States.

”

While we certainly don't yet have all the answers we wish we had for prevention of VHL tumors, we do know some, and the most important one of all is early diagnosis. Knowing you are at risk, you have the opportunity to take the first steps. Failure to go for testing can have disastrous consequences -- both psychologically and physically.

In sports, you can't hope to win the game if you don't know the rules. Find out what game you're playing and work with your medical team to maintain your health!!!

Other things you can do:

- Get *real information* on your condition; don't let fears rule your actions.

- Stop smoking yourself, and among others in your home and workplace to reduce your exposure to second-hand smoke. Don't use smokeless tobacco either.

- Limit consumption of alcohol as well as smoked and cured foods.

- Eat a variety of fruits and vegetables daily, but minimize your exposure to insecticides. Wash fruits and vegetables carefully, and consider buying organically grown.

- Reduce the amount of meat in your diet; eat a diet based predominantly on plants.

- Increase your intake of high-fiber foods. Include some soy products at least once or twice a week.

- Maintain a desirable weight; engage in moderate exercise.

Diet recommendations are from the American Cancer Society. Case histories are compiled from real people who are members of the VHL Family Alliance; identities have been obscured.

Reeve pleads for Brain Research and Insurance Reform

-- adapted from an article by Judith Havemann, *The Washington Post*, April 1996

In better days, Reeve strode into a Capitol Hill hearing room exuding the charm and confidence that made him one of Hollywood's favorite leading men -- the unmistakable Superman speaking in behalf of funding for the arts.

Now Christopher Reeve sits in a wheelchair in his suburban New York home hooked up to a respirator, his powerful voice reduced to short gasps of eight to 10 words at a time. Paralyzed in a horseback-riding accident in May and facing a lifetime of disability, he has become one of the millions of Americans trying to plan uncertain futures around the fine print of health insurance policies.

Reeve does not suggest that his financial picture is that of the average person, and his ability to negotiate with the web of insurers now involved in his personal crisis is far greater than most. But his injury has brought home to his family the fragile nature of fortunes and futures based on expectations of health and continued employment. Reeve has approached his problem in typical fashion. He is fighting back, using his political clout to position himself as an activist for changing national policy.

"A year or two ago we seemed to be very well off," said Reeve, talking into a speaker phone from his home in Pound Ridge, N.Y., where he lives with his wife and his 3-year-old son, Will. Now, "the picture has changed. And there are so many other people who are in this situation whose positions are so much worse."

Reeve's principal health insurance -- bought 20 years ago, through the American Federation of Television and Radio Artists, when his career was blossoming -- is rapidly running out. Care for his catastrophic injury will cost about \$400,000 a year, an extraordinary cost faced by relatively few Americans. His main policy (through an insurer he declines to name) has a \$1.2 million lifetime cap on payments for catastrophic injury, slightly more than the typical \$1 million, industry statistics show. This gives Reeve about three years of coverage.

Business groups call such caps "regrettable but necessary," designed to keep insurance costs from skyrocketing. About 70% of the nation's health insurance policies have lifetime caps, often never noticed by the policyholder until disaster strikes.

Reeve's wife, Dana, described discovering their policy's cap as a "one-two punch. When I really looked in depth at the policy, to be quite honest, it was . . . as horrifying to me as when Chris first had his accident. It was one of the worst days, when I realized

what our financial situation would be and what Chris's care would cost."

Reeve is writing to every U.S. Senator and endorsing legislation by Sen. James M. Jeffords (R-Vt.) that would change the lifetime cap. Jeffords sought him out for the help, knowing of Reeve's situation. Under the amendment, no insurance company would be allowed to set a cap of less than \$10 million.

Debate over the issue has been bitter and -- up to now -- largely hidden in the grinding health reform fight on Capitol Hill. The amendment would be added to the health care reform measure being pushed by Sen. Nancy Landon Kassebaum (R-Kan.) and Sen. Edward M. Kennedy (D-Mass.), which would stop insurance companies from dropping people when they change jobs or denying coverage for pre-existing conditions. That measure unanimously passed the Senate Labor and Human Relations Committee in August, but it has been stalled ever since by secret parliamentary tactics.

The basic Kassebaum bill has run headlong into opposition from some powerful insurance companies. Amendments like the one endorsed by Reeve have encountered even more visceral reaction from the larger business community.

James Klein, president of the Association of Private Pension and Welfare Plans, said Reeve presented a "tremendously compelling fact situation" and expressed profound sympathy for Reeve's "extraordinary tragedy." But, he said "limits are a regrettable but necessary part of many health plans. Without them, many employers -- those who are voluntarily providing health care benefits to their employees -- wouldn't be able to provide health care coverage at all."

Reeve said dropping the lifetime limits on medical payments and spreading the cost of the few catastrophic cases across all policyholders would increase a typical policy's cost about \$8 a year, a figure disputed by some in the industry. Klein said cost increases would vary depending on the benefits provided by a company but could be financially ruinous.

Once these insurance caps are reached, taxpayers often pick up the liability through the federal Medicaid program. About one-third of total Medicaid costs go to the disabled, who compose less than 15% of those enrolled. And a disproportionate share of those resources go to a relative handful of severely incapacitated individuals whose intensive care costs hundreds of thousands of dollars a year.

"Their costs are so high that if left unchecked they will bankrupt the country," Reeve said.

Personally, he said, he had sufficient resources not to need Medicaid. But, his voice bristling, he said "you're mistaken if you think" he could support costs of \$400,000 a year "unless I find work."

Reeve has no memory of falling off his horse last Memorial Day weekend at a riding exhibition in Culpepper, Virginia. He never has flashbacks or nightmares.

Unconscious for four days, he woke up to learn he was paralyzed and would need an operation to stabilize his head and spine as soon as his lungs were clear of pneumonia.

He broke his first two cervical vertebrae, crushing his spinal cord where it exits from his brain, in effect cutting off his body from his mind.

Only gradually did he become aware of what his family already knew: in addition to the massive injuries, he didn't have enough insurance to pay for his medical care. He needs nurses 24 hours a day, aides to lift him in and out of his wheelchair, and therapists to keep his body from atrophying.

"I'm going to go back to work," he said repeatedly. He hopes to be able to take advantage of some directing offers next fall, and he could command substantial fees for speaking around the country.

Since the accident, Reeve said, he has "experienced some very slow recovery. I have increased sensation in my left leg and increased movement in my shoulders."

Last November 2, he said, "I was not able to take one breath without the ventilator, but as recently as last week I was able to be off the ventilator for 90 minutes, and I think that will increase."

Two weeks before Christmas, Reeve was discharged from the Kessler Institute for Rehabilitation in New Jersey. But his health remains precarious. He was rushed to Northern Westchester Medical Center last month with an erratic blood pressure condition caused, in this case, by a urinary tract infection.

He still depends on the ventilator to breathe, and it occasionally stops. "If there were a malfunction in the ventilator, I would not survive unless there were somebody there."

His insurance company rules important aspects of his life. It refused, for example, to let him leave the Kessler Rehabilitation Hospital overnight to spend Thanksgiving with his family. But he understands the insurance companies better than most; his grandfather was board chairman of Prudential Insurance Company. "I remember him saying that they felt an obligation to their customers from the cradle to the grave," he said.

Reeve appears absolutely confident that a cure will be found for spinal cord injuries in ten years. Researchers are working to regenerate crushed nerves, possibly allowing victims to walk again. A \$40 million-

a-year government investment in research, Reeve said, could save billions in Medicaid and Medicare costs.

It is the "proper humanitarian and economic thing to do," he said. "All people in my situation are desperate not to be a burden to their families, to government, or to the insurance companies."

Brain Awareness Week

May 12-18 marked Brain Awareness Week, a national public information campaign created by the Dana Alliance for Brain Initiatives to unite scientific institutions, professional organizations, advocacy groups and public officials under the umbrella theme, *Brain Research for the Life of Your Mind*. This is a common effort to expand research on the treatment, cure, and prevention of brain diseases and disorders that affect more than 50 million Americans. The VHL Family Alliance is a partner and participated in events on Capitol Hill in May.

Chairman, Christopher Reeve, representing the American Paralysis Association, has given this movement a level of visibility which it would never have achieved without his sponsorship.

The partners in Brain Awareness Week include 15 government entities, including 11 of the National Institutes of Health, the Library of Congress, and over 125 scientific, educational, and patient outreach groups. A website is available at

<http://www.dana.org/brainweek>

As part of the campaign, people of all ages across the country were able to visit brain research laboratories, take a brain quiz, see exhibits on the brain, hear lectures from prominent scientists, find information about the brain through an Internet web site, and participate in many other national and local activities.

Brain Quiz

Which of the following are disorders of the brain?

- ☐ a. Cerebral Palsy, Epilepsy, Paralysis, Spinal Cord injury, Multiple Sclerosis, VHL spinal lesions.
- ☐ b. Schizophrenia, Depression, Alcohol Addiction, Anxiety Disorders
- ☐ c. Autism, Anorexia, Learning Disabilities, Rett Syndrome, Dyslexia, Migraines
- ☐ d. Alzheimer's Tourette Syndrome, Parkinson's, VHL and other Brain Tumors, Lou Gehrig's Disease.
- ☐ e. All of the above.

Answer: e. All of the above. Brain research demonstrates that disorders as different as addiction, learning disabilities, and Lou Gehrig's disease all have their roots in the brain. Brain-related disorders range from wide-spread conditions such as pain, which affects virtually everyone, to rare disorders such as Williams Syndrome, a cause of mental retardation in one in 20,000 children, or VHL, which causes tumors in one in 32,000 people worldwide. Every American will be affected at some point in his or her life, either personally, or with a family member's struggle with a brain disorder.

Surveys as a Learning Tool

-- Joyce Graff, Editor

At the Boston meeting we distributed some on-site questionnaires to test what we might be able to learn from each other, and what useful information we might be able to provide to ourselves and our physicians. Our goal is, as ever, to work toward improvements in diagnosis, treatment, and quality of life for all of us. Here are the results of the Boston On-Site Survey.

The goal of this section was to learn what was working best in reaching VHL families. Respondents were meeting attendees, not all of whom have VHL themselves.

How did you first hear about us? 12% heard about us from a doctor, 18% from a friend, 45% from a relative, 9% from a brochure, 9% from newspaper or magazine, 6% from the internet.

How long did you wait before contacting us? 46% waited a few days, 23% waited a few weeks, 23% waited months, 9% waited longer than that.

How did you first reach out to us? 69% reached out by telephone, 27% by letter, 4% by e-mail. We have seen a marked increase in internet contacts over the last year, and will be interested to compare the next round of results.

Then we asked a number of questions specifically to people with VHL. We wanted to compile their experiences of gaining a diagnosis. Of people with VHL, 37 people responded, ranging in age from 19 to 59 (average age 38).

How did you first learn you had VHL? (a) It runs in the family, so I knew I was at risk (37%); (b) Another family member was diagnosed so they checked me too (30%); (c) I was the first person in my family to get a diagnosis of VHL (30%).

How old were you when you had your first symptoms? Ages ranged from 9 to 59 (with a statistical mean of 19.2 years). 14% of those responding had no symptoms.

How old were you when you were diagnosed? Age at diagnosis ranged from 9 to 59 (mean=22). One man was screened until he was 50 years old, was told he could stop screening since he was too old to get VHL, and was subsequently diagnosed at age 59 with kidney cancer. One person still has no clear diagnosis though she has had multiple spinal tumors.

In 68% of the cases, diagnosis was made because of symptoms, while 31% of these people were diagnosed through asymptomatic testing.

What physician did you first go to with your symptoms? primary care physician (33%), neurologist or neurosurgeon (22%), ophthalmologist (27%), urologist (11%).

At first the doctors thought it was... (a) VHL (68%); (b) hypochondria (9%), (c) multiple sclerosis (4%), (d) sporadic brain tumor (18%). Other early misdiagnoses included random migraines, seizure disorder of unknown origin, sporadic pheo, polycystic kidney disease, dual retinal hemangioma (not VHL), and carpal tunnel syndrome.

Testing was ordered by the... primary care physician (43%), pediatrician (3%), ophthalmologist (7%), geneticist (7%), or only on the patient's own insistence (2%).

These answers tend to indicate that about 68% of the time the system worked reasonably well. It also gives us some clues as to how we might provide some additional information to physicians which would shrink that 32%. The system will not work perfectly until there is a simple test for VHL, but some additional information on differential diagnoses could help physicians move more rapidly from these first impressions to a diagnosis of VHL.

Of the 25 women responding, 24% reported having cysts and 16% some sort of tumor (fibroids or other) in the uterine area. Cysts and tumors are so prevalent among women that it is difficult to draw any conclusions from these replies.

Of the 11 men responding, 78% reported having epididymal cystadenomas (epi cysts). While 11 is too small a sample to be statistically significant, this is a much higher percentage of penetration of epi cysts than reported in any of the literature. 44% of these men found them in their teens, 33% in their 20's, and one in his 50's. Half first discovered them himself, and half were first found by the physician (most of these at NIH). None of these men discussed this issue with a relative, all went to a doctor with their concerns. Half were dissatisfied with the advice given them by the doctor. 80% of those with epi cysts reported no blockages or other problems with them, one was infertile due to epi cysts. We are adding some information on epi cysts to the new handbook which we hope will be more helpful.

We hope that you will participate in surveys, and that you will share with us your opinions about the surveys themselves and how we can improve them. We are always interested in your feedback, positive and negative. Your participation is encouraged, but of course is always voluntary. The greater the number of people participating, the more meaningful the results.

□

1996 RESEARCH QUESTIONNAIRE

In an effort to further research, the VHL Family Alliance is compiling a database of patients who are affected with VHL.

From time to time the VHL Family Alliance may receive requests for names of families willing to participate in research. Only bona fide research projects will be considered that have met with the approval of our Research Management Committee.

Are you willing to be contacted directly by VHLFA approved medical researchers who are studying VHL?

☐ Yes ☐ No ☐ Ask me each time

If you mark "NO", the information which you provide by completing the attached form will only be released in summaries which do not identify you by name in any way. If you mark "YES" you may or may not be contacted. If contacted, you have NO obligation to respond or to participate in any way.

Are you willing to be contacted by medical researchers who are seeking tissue or blood samples?

☐ Yes ☐ No ☐ Ask me each time

Signature: _____

Printed Name: _____

Parent or guardian of a minor child:

Signature: _____

Printed Name: _____

Witness: _____

Printed Name: _____

Date: _____

Your participation is totally **voluntary**. You can ask to have this form returned to you at any time. You will never be pressured to do anything or to participate in any research unless you so choose. Please complete one form for each person affected by VHL. You may be asked to update this information annually. Your comments are most welcome.

Thank you for your participation!

RESEARCH QUESTIONNAIRE

Name: _____
Last First Middle

Address: _____
Street

City State Zip

Telephone: _____
Home Work

Birthdate: _____ Sex: ☐ Male ☐ Female

Ethnicity: ☐ American Indian/Alaskan native
☐ Asian/Pacific Islander
☐ African or African American, not of Hispanic origin
☐ Hispanic
☐ Caucasian/White, not of Hispanic origin

Have you participated in any studies at the National Institutes of Health in Bethesda, Maryland?

☐ Yes ☐ No

FAMILY HISTORY:

Are your parents affected by VHL?

Mother: ☐ Yes ☐ No ☐ Maybe ☐ Probably ☐ Unknown

Father: ☐ Yes ☐ No ☐ Maybe ☐ Probably ☐ Unknown

Do you have brothers or sisters?

☐ Yes. Number of brothers _____ Number of sisters _____

Have all siblings been evaluated for VHL? ☐ Yes ☐ No

Are they affected by VHL? If yes, how many? _____ brothers _____ sisters

Do you have children?

☐ Yes If so, how many? _____ ☐ No

How many of those children were diagnosed with (or are believed to have) VHL?

____ sons ____ daughters

Do you have other relatives affected by VHL?

☐ Yes ☐ No ☐ Maybe ☐ Probably ☐ Unknown

Number Yes ____ # No ____ # Maybe ____ # Probably ____ # Unknown ____

Have any of your family members completed a questionnaire like this for VHLFA?

☐ Yes ☐ No Name(s) _____

May we send you questionnaires for your family?

☐ Yes ☐ No How many? _____

DNA TESTING:

Are you interested in DNA testing for your family?

☐ Yes ☐ No

Has the mutation in the VHL gene been found for your family?

☐ Yes ☐ No

If yes, what is the nucleotide change or name of the mutation?

Which laboratory found the mutation?

How do you think DNA testing can benefit your family?

- ☐ To tell us who needs testing and who does not
☐ To predict which VHL affects are more likely to occur than others
☐ To assist in child-bearing decisions
☐ To perform pre-natal testing of embryos
☐ To perform pre-implantation testing of zygotes

☐ Other: _____

Would you be willing to pay \$1000 to find the mutation in your family?

☐ Yes ☐ No ☐ Other _____

This would then be a "road map" which could be used to simplify testing for other family members. Estimated cost for additional people would be \$250-300 each.

MEDICAL HISTORY:

Von Hippel-Lindau disease is a disease which can have a variety of symptoms. Please check as accurately as possible the symptoms which apply to you. **N/E** stands for "not evaluated", that is if you have not had screening for that particular body part. Under the **Surgery** column, please list the date(s) of surgery. The word *lesion* is used to describe angiomas, hemangioblastomas, and tumors.

Age at first symptoms: _____ Age when first diagnosed: _____

First Symptoms: _____

EYES	YES	NO	SURGERY	N/E
Retinal lesions	_____	_____	_____	_____
Other _____	_____	_____	_____	_____
What is the status of the vision in your				
Left Eye <input type="checkbox"/> Good vision <input type="checkbox"/> Partial vision <input type="checkbox"/> Blind <input type="checkbox"/> Enucleated				
Right Eye <input type="checkbox"/> Good vision <input type="checkbox"/> Partial vision <input type="checkbox"/> Blind <input type="checkbox"/> Enucleated				
BRAIN	YES	NO	SURGERY	N/E
Cerebellum lesions	_____	_____	_____	_____
Brain stem lesions	_____	_____	_____	_____
Other (e.g. pituitary)	_____	_____	_____	_____
SPINAL CORD	YES	NO	SURGERY	N/E
Lesions inside cord	_____	_____	_____	_____
Lesions outside cord	_____	_____	_____	_____
Other _____	_____	_____	_____	_____

KIDNEYS	YES	NO	SURGERY	N/E
Cysts	_____	_____	_____	_____
Renal Cell Carcinoma	_____	_____	_____	_____
Other _____	_____	_____	_____	_____
PANCREAS	YES	NO	SURGERY	N/E
Cysts	_____	_____	_____	_____
Lesions	_____	_____	_____	_____
Other _____	_____	_____	_____	_____
ADRENAL GLANDS	YES	NO	SURGERY	N/E
Lesions	_____	_____	_____	_____
High blood pressure	_____	_____	_____	_____
Other _____	_____	_____	_____	_____
EPIDIDYMS (Gland next to the Testes)				
(Men Only)	YES	NO	SURGERY	N/E
Cystadenomas	_____	_____	_____	_____
Other _____	_____	_____	_____	_____
FALLOPIAN TUBES or Broad Ligament				
(Women Only)	YES	NO	SURGERY	N/E
Cystadenomas	_____	_____	_____	_____
Other _____	_____	_____	_____	_____
Childbearing (Women only)				
How children have you had? (live births) _____		How many miscarriages? _____		
HEARING CHANGES	YES	NO	SURGERY	N/E
Lesion or mass	_____	_____	_____	_____
Frequent ringing in ears	_____	_____		
Any other changes in hearing, please describe:				

Left side _____

Right side _____

OTHER things you think may be related to VHL...please explain below.

- | | | |
|--|--|--|
| <input type="checkbox"/> Hormonal problems? | <input type="checkbox"/> Sweating attacks? | <input type="checkbox"/> Diabetes? |
| <input type="checkbox"/> Digestive problems? | <input type="checkbox"/> Eating disorders? | <input type="checkbox"/> Headaches? |
| <input type="checkbox"/> Bloating of abdomen? | <input type="checkbox"/> Panic attacks? | <input type="checkbox"/> Spiking blood pressure? |
| <input type="checkbox"/> Hiccups? | <input type="checkbox"/> Sneezing? | <input type="checkbox"/> Allergies? |
| <input type="checkbox"/> Prematurely gray hair? (what age? _____) | | |
| <input type="checkbox"/> Blistering following surgery? | | |
| <input type="checkbox"/> Skin lesions | | |
| <input type="checkbox"/> Fatty tumors | <input type="checkbox"/> Raised moles | <input type="checkbox"/> Flat brown spots (state size) _____ |
| <input type="checkbox"/> Other things you feel may be related to VHL, please describe: | | |

TREATMENTS:

Have you ever been treated with Gamma Knife, Lineac, or other Stereotactic Radiation? ☐ Yes ☐ No

Have you ever been treated with kidney dialysis or transplant? ☐ Yes ☐ No

If yes, we will send you a supplementary questionnaire on these topics.

OTHER MEDICATIONS:

Have you ever undergone any of the following therapies, or another course of medication or radiation which is not listed here?

- ☐ Interferon ☐ Interleukin ☐ Thalidomide

(which kind?) _____

- ☐ Conventional radiation therapy? Where? _____

☐ Other _____

What was the goal of the treatment?

When and for how long did you undergo this treatment? (or
“continuing”) _____

What drug(s) were used for post-treatment management?

What post-treatment effects did you experience?

How long did the post-treatment recovery period last?

If you encountered specific problems, please detail them (add paper as needed)

At this point you are ____ months post-treatment. Would you say that this treatment was successful? ☐
Yes ☐ No Would you recommend it to others? _____

Comments on this questionnaire? Are there things you would change? Add? Delete?

What would you like to learn? In next year’s questionnaire, what would you like us to ask the group?

Thank you!

Please send your completed questionnaire to

VHL Family Alliance

Research Committee, William C. Dickson, Chair

10613 Creamcup Lane

Great Falls, VA 22066

703-759-3665; Fax: 703-759-7992; e-mail: vhlres@usa.pipeline.com

Living with a Rare Disorder . . .

from the National Organization for Rare Disorders, Resourceful Steps workshop, 30 September 1995.

. . . can be challenging. In addition to the physical discomforts and limitations, there is the added burden of dealing with most people's unfamiliarity with your illness. Over 5,000 rare diseases collectively affect 20 million Americans. Yet, most patients remain misdiagnosed or undiagnosed for a long period of time -- many remain undiagnosed for six years or more.

Rare disorders affect less than one out of every 100 people.. VHL, for example, is estimated to affect one in 32,000 people. Diseases like VHL have become known as "orphan diseases" and the drugs developed to treat them are called orphan drugs.

In 1973 only ten products were approved that would have fallen under today's definition of orphan drug. This was because the market was too small for pharmaceutical companies to consider it a worthwhile investment. In 1983, the situation changed when Congress passed the Orphan Drug Act. NORD was instrumental in lobbying for this act, which enables companies willing to develop orphan drugs to recoup the cost of researching and developing them by providing tax incentives and sole marketing rights. Since the Orphan Drug Act was introduced, over one hundred orphan drugs has been approved and dozens more are being researched.

If a drug therapy does exist and is available, it may well be very expensive. Then there are laboratory test costs, specialized equipment costs, caregiver expenses, and travel expenses to distant medical facilities. Even with adequate health insurance in place, co-payment expenses can be overwhelming. But the biggest cost of all is often the unseen toll that dealing with chronic illness takes on patients and their families and friends. When the illness is a rare disorder, there is the added difficulty of tracking down adequate information and hard-to-find support groups.

When you are looking for information about and referrals to sources of help, you will need a great deal of determination, perseverance and, most of all, hope. The following "Five Steps toward the Light" are meant to guide and advise you as you begin your search.

Five Steps Towards the Light

1. When you go to the doctor, write down any information that you want to share, including questions and observations. Be assertive -- patients should not feel intimidated by their physicians. Solid data that can be substantiated with published facts and/or physical findings in the patient can be medically important. If your doctor repeatedly refuses to learn from you, consider finding another physician.

2. Get hold of your medical records -- you can't remove them from a facility but you can get copies made and take them with you. This is particularly

important if you have a rare disorder and are seeking medical care from a new provider. Try to get a second opinion from a specialized source, such as a major medical center or teaching facility.

3. Be aware that rare diseases can be as frustrating to the physician as to the patient, possibly leading the doctor to recommend psychiatric help. Sometimes, even the patient's family and friends have difficulty believing the patient is really suffering. Find a support group where you can get emotional and moral support as well as information about your illness.

When you start calling agencies and organizations for help and support, keep your information organized, accessible, and legible. Write a list of your questions and concerns so you will not have to gather your thoughts while on the phone. Make a note of responses to your questions, as well as the names of the people and agencies you contact, and their phone numbers. If the person you talk to seems rude or insensitive, try to stay calm. Don't forget that everybody has bad days!

4. Keep in mind the many local sources of help available from charitable, religious, and civic organizations. Also, the reference section of your local library may be able to find you useful referrals. Many national organizations have local chapters that may be listed in your telephone book. If there is no local chapter, and no toll-free number for the national group, you may prefer to contact them in writing. Make sure to write legibly, in block capitals if possible, and always include your complete name, mailing address, zip code and phone number (including the area code) in the body of the letter.

5. Always be hopeful. Be persistent, but not unpleasant. Have the determination to keep on trying. **You** can make a difference; better yet, you may even make **the** difference!

Remember -- every journey begins with a single step!

National Organization for Rare Disorders, New Fairfield, Connecticut, 1-800-999-NORD. For VHL, the VHL Family Alliance will be happy to answer your questions by phone, fax, or E-mail, and can refer you or your physician to sources of second opinions, 1-800-767-4VHL. For local assistance with counseling, support groups, transportation, and volunteers, consult your local United Way, Cancer Society, Brain Tumor or Kidney support organizations, and the Well Spouse Foundation. Counselors skilled in working with people with chronic illness may be found through Diabetes or Kidney organizations, children's hospitals, or large medical centers.

VHL and the Hawaii Family

-- with Rodney and Vanessa B., Hawaii

Long before they became the subject of genetic research in the early 1980s, Rodney B's Hawaii family would talk among themselves about why so many of them had cancer. In a sense, learning that it was a genetic disease helped to make some sense of it, and to give them ways of fighting back.

At the turn of the century, their ancestor, Lazarus R., brought his young family to Hawaii after a hurricane devastated Puerto Rico. Lazarus died en route, but his widow and two little children ended up on Kauai. The little girl, Helen, had the flawed gene. She had three husbands and some 15 children. Her brother Frank, who also had VHL, had eight children who lived to adulthood.

Helen's descendants, now scattered across the Islands and the Mainland, number some 260 or more members over six generations.

"I've always said, 'Ah, the family curse,'" Says Roxane M., another family member. "But David T., another cousin, says, 'It's no curse, it's a blessing.' If it were not for VHL, I would never have met David. I would never have started a family newsletter. Now I want a family reunion."

Members of this extended family have shown extraordinary grace, and grown closer, as they fight VHL. They support one another, encourage each other to get tested for the gene, and grieve when the news is bad. Some worry, others try to forget.

"I talked to my brother and he's just glad he doesn't have it," says Rodney. "He told me once he's afraid if he had it, he would be devastated. But you do what you have to do. When I find out others have problems, it bother me more than my own. I don't want them to suffer."

Kidney cancer has been a particular problem in this family. Rodney has had eye and brain tumors, but kidney cancer has been his single biggest issue. In his twenties a series of large cancerous tumors led to removal of both kidneys. He had a transplant which failed after six years. At age 40, Rodney has dealt with years on dialysis and two hip replacements, complications from long-term use of steroids. His is a very tough story of life with VHL. "There's some days you cry. But I think God gave me grace in deal-ing with it," says Rodney, staring with blurred vision at the blaze of bouganvillea on his townhouse lanai.

His wife Vanessa has been a wonderful partner. They travel together with their one-year-old daughter, no small undertaking for someone on dialysis. But they have learned to organize around dialysis and life's other challenges, medical and otherwise, with good humor and determination.

Roxane watched her father die at age 49, also of kidney cancer. "My Dad told my husband, 'Don't have

kids,'" says Roxane. "We talked about it, but I decided I was going to have kids whether or not they might have VHL. You can die from other things too."

Roxane knew that, like everyone in her family, she was at 50% risk of having VHL, and has gone every 3 years for the last two decades for thorough screening which includes an MRI brain scan, gynecological check, eye exam, and kidney ultrasound.

"What scares me is my children are at risk," Roxane said in an interview last year. "But you have to go on about life. You can't worry about this thing. One of my aunts doesn't have it, and she was just diagnosed with breast cancer. If it's not one thing, it's something else. You've got to remain positive."

Since the discovery of the gene in 1993 there is now a blood test to screen for VHL. A grant from Kapiolani Medical Center is making the test available free to family members. Already 34 people have been tested.

"It's a cancer-susceptibility disease," explains Dr. Y. Edward (Ted) Hsia, the geneticist who has pushed national researchers to isolate the gene and find better medical treatments for people with VHL. "It's a tumor-suppressor gene. When the gene is working properly, it prevents cancers from forming. When it doesn't work right, it allows tumors to form." People with the flawed VHL gene get more tumors more often than people in the general population.

Annual screening could save a wealth of human suffering and medical costs, Hsia says. Yet medical insurers are sometimes loathe to pay for such screening. DNA testing can help to determine who is and is not at risk. After living for 35 years believing she was at risk for VHL, Roxane recently learned that she does not have the gene. "It's a great relief in some ways, especially knowing now that my children are not at risk for VHL. But it's also a tremendous adjustment. In a sense I have to redefine who I am. I feel almost guilty for not having the gene, almost as if I had lost my place in the family." Roxane's feelings are quite normal, a reaction known as "survivor guilt," typical of unaffected children in a family united by a common problem.

In a little Hawaiian country town, David T. was Kauai's first successful kidney transplant, which has lasted him nearly eighteen years. He receives full military disability, and serves as pastor to a tiny Missionary Baptist Church. David was 22 when doctors first diagnosed the VHL tumors in his kidney, eyes, and brain stem. Now his 17-year-old son has been diagnosed with VHL too.

"With my boy, we knew he had a 50-50 chance of having it. He grew up in this house. He was always aware of VHL."

"When we got the news he had these growths in his kidney and pancreas. Initially we were swept away. Just like me, he had no symptoms, so we had

no warning. But he has a really good attitude.

"I told him, 'You can't let this thing take away from your quality of life. You can have a good life. We just have to manage the disease.' And we are learning more every day about better management and better treatment."

Although some of the family members are bitter and angry about the disease that has taken some lives and left others with disabilities, both Rodney and David express optimism.

"Tomorrow isn't guaranteed to any of us," says David. "You have to be wise and appreciative of today."

Rodney, who spends several hours three times a week on dialysis and keeps hoping for another transplant, has similar sentiments. "When people go through a lot, their mercy grows. When I see someone hurting, my heart goes out to them. When they're having pain, I understand."

Thanks to Beverly Creamer, Honolulu Advertiser, for her assistance in the preparation of this article

Tylenol and Alcohol: A Dangerous Cocktail

-- adapted from an article by Sidney M. Wolfe

So many of us take over-the-counter remedies for the lesser aches and pains of VHL and life in general, we wanted to warn people about possible liver damage from non-aspirin pain relievers when you drink alcohol. -- Editor

A 39-year-old man who normally drank two to four glasses of wine at dinner ran into trouble when he began taking Tylenol, the principal ingredient of which is acetaminophen, a non-aspirin pain-reliever, in the recommended dose for the flu. The man developed liver failure and required an organ transplant to save his life.

He sued the manufacturer and won a multi-million-dollar judgment, which was upheld on appeal. Since then, additional lawsuits have been filed, alleging that the manufacturer did not adequately warn about the potential harm when acetaminophen and alcohol are mixed.

This connection is not new. Case reports have appeared in medical journals since 1977 about liver damage from the mixture of acetaminophen and alcohol. There are 94 known liver damage cases. In September 1995, researchers reported in the liver-specialty journal *Hepatology*, 67 more cases of serious liver damage linked to acetaminophen taken in usual therapeutic doses for adults (not overdoses taken in suicide attempts) and alcohol. This brings the total reported cases to 161, including 32 deaths.

Sixty-four percent of the new cases were considered alcoholic; however, 35 percent drank less than the equivalent of four glasses of wine, or three mixed drinks containing hard liquor per day.

While the U.S. Food & Drug Administration (FDA) has proposed rules requiring an alcohol warning on all non-prescription pain relievers, not just those containing acetaminophen, it now appears that these rules will not be promulgated before mid-1996. Some manufacturers have already voluntarily added the warning: *If you generally consume three or more alcohol-containing drinks per day, you should consult your physician for when and how you should take Tylenol and other pain relievers.*

Public Citizen Health Research Group finds this warning totally inadequate. It does not inform consumers that the potential problem is liver toxicity, or that liver toxicity is potentially life-threatening.

What You Can Do

If you drink alcohol, but less than four glasses of wine or three mixed drinks a day, use acetaminophen in the lowest possible dose to relieve your symptoms -- in no event more than four extra-strength or six regular-strength tablets daily -- and stop taking the drug as soon as your symptoms subside.

If you are using acetaminophen to treat cold or flu symptoms and regularly use alcohol, stop the alcohol until you are over the cold or flu, as it can aggravate cold and flu symptoms. *Remember that many liquid cold remedies contain alcohol and count as shots of whiskey.*

If you use acetaminophen regularly to control pain and use alcohol, keep your dose of acetaminophen as low as possible and moderate your alcohol intake.

The most common complaints reported by those experiencing the acetaminophen-alcohol interaction were nausea, vomiting, abdominal distress or pain. These are very non-specific complaints and are symptoms common to many mild disorders. The only sure way to detect liver toxicity is with a blood test ordered by your doctor.

Heavy drinkers should consult a physician before taking any pain reliever on a regular basis.

Combining heavy drinking with aspirin or ibuprofen (such as Advil) may promote gastrointestinal bleeding. Acetaminophen (such as Tylenol) and heavy drinking may promote liver disease.

Sidney M. Wolfe, Editor, *Worst Pills/Best Pills News*, 2:3, March 1996, Public Citizen Health Research Group, Washington, D.C.

What is Moderation?

Moderation is defined as no more than one drink per day for women and no more than two drinks per day for men.

Count as a drink--

- 12 ounces of regular beer (150 calories)
- 5 ounces of wine (100 calories)
- 1.5 ounces of 80-proof distilled spirits (100 calories)

-- Nutrition and Your Health: Dietary Guidelines, 4th edition, 1995, U.S. Department of Agriculture, Home & Garden Bulletin #232.

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Committee and Board changes

Peggy Graham, Patti Kohlen, and Susan Warnick complete their terms on the Board of Directors this year. All three have played key roles in the founding and development of the VHLFA. We thank them very much for their excellent service to the VHL Family Alliance, and welcome their continued participation. We are pleased to welcome a number of new participants in the running of the VHL Family Alliance. Board elections will be held at the Annual Meeting in Hawaii.

Melissa Minster, chair of the Delaware Valley Chapter, is nominated to the Board of Directors. Altheada Johnson is nominated as Chairman of the Board, replacing Susan Warnick as our Chief Executive Officer. Joyce Graff is nominated for re-election to the Board and to continue as Chairman of the Alliance, our Chief Operating Officer.

The Public Relations Committee will be chaired by Micheline Massé of Arizona. Joining her in that effort is Cary Schwanitz of Texas.

Peggy Graham turns over the reins of the Professional Education Committee to Donna Russo, genetic counselor at Columbia-Presbyterian Hospital in New York City.

Susan Warnick will continue as Chair of the Clinical Care Centers Committee. There are positions open with this committee.

Renée Rosado of California joins us as Vice Chair of the Fund-Raising Committee, with a focus on supporting research.

Melissa Minster Nominated to the Board

A fourth generation jeweler who has been working in the family business almost since birth, Melissa is taking the business in a new direction with a line of Minster Design Jewelry, designed and manufactured on location. One of her designs is the VHL Team-work Pin.

In 1995 she joined the Board of Directors of the Newark branch of the American Cancer Society. She has served on various committees throughout the year.

Melissa is the youngest of three siblings diagnosed with VHL, which was inherited from their father. As a family they have been dealing with VHL since the 1960's. Until joining the VHL Family Alliance they had very few true facts about VHL.

Melissa has been actively involved in the VHL Family Alliance since joining in 1993. "Working with the VHL Family Alliance has given us a sense of power and belonging in fighting this disease. With the increased attention and research focused on von Hippel-Lindau we can all live longer and healthier happy lives."

Melissa Minster, Delaware

Happenings

Women and Genetics in Contemporary Society, May 16-19, Zanesville, Ohio.

Altheada Johnson represented VHLFA, participating as a presenter on two panels: Cancer Susceptibility and Disability Concerns. The focus of the conference was to control the potential dangers inherent in the discovery and use of genetic information, while taking advantage of the potential benefits of this information. Altheada takes the position that this technology is a good thing, and that with reasonable safeguards we should be able to reap significant benefits.

The Responsibility of Oversight in Genetic Research and Technology Development, May 9-10, Boston, Massachusetts, sponsored by Public Responsibility in Medicine & Research. Joyce Graff represented VHLFA on a panel, What consumers of genetic services expect from the genetics research establishment. She made the point that consumers want to be *partners*, not "subjects", in research.

Cancer in Families, 2nd International VHL Symposium, Honolulu, June 17-21, 1996. Dr. Y. Edward Hsia and the Department of Medical Genetics at Kapiolani Hospital in Honolulu and the VHL Family Alliance are co-sponsoring this year's conference on VHL. We look forward to reporting this exciting program in the September issue.

Workshop '97 on MEN2 and VHL. Dr. Cornelis Lips in Utrecht will be sponsoring a Workshop June 25-28, 1997, in Noordwijkerhout, a beautiful congress center near Amsterdam in the Netherlands. The Sixth International Workshop of Multiple Endocrine Neoplasia will next year also include von Hippel-Lindau disease. Delegates are invited from a wide range of disciplines, and there will be some involvement of the family support groups for these two conditions. For more information please contact Cornelis J. M. Lips, M.D., Department of Internal Medicine, University Hospital Utrecht, Heidelberglaan 100, 3584 CX Utrecht, The Netherlands. Phone +31 (30) 2 509 111; Fax: +31 (30) 2 518 328.

The 32 participants of the California Chapter's annual meeting on April 13 are gathered around the Gamma Knife unit at the University of California in San Francisco where they received a tour of the unit and were updated on the latest procedures from UCSF's Gamma Knife neurosurgeon, Dr. Mike McDermott. This very successful meeting ended with the election of the Chapter's Board of Directors and Officers.

The Murray Foundation and the VHL Family Alliance presented matching checks to the Massachusetts General Hospital April 14 in Boston, to kick off "our" research project, The Crystal Structure of the VHL Protein. Left to right: Scott Murray, Joyce Graff, J. Patrick Murray, Jane Claflin (Mrs. R. Morton Claflin) of the MGH Board, accepting the checks on behalf of Dr. Diana Griffith.(right).

VHL Family Forum

Newsletter of the VHL Family Alliance
171 Clinton Road
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