VHL Family Forum

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VHL Disease in France

by Stéphane Richard, M.D., Ph.D., and François Resche, M.D., Paris

For generations in France, as in other countries, von Hippel-Lindau disease was seen as a rare condition of cerebellar and retinal tumors. Visceral manifestations, when they were considered at all, were thought of as curiosities, without significance in the course of the disease. Today, due to progress in brain imaging, microsurgery, and post-operative care, neurosurgical manifestations of the disease are comparatively easy to manage. Persons affected with VHL disease tend to live longer, and diagnosis is more accurate. This has led to the realization that VHL is more prevalent than previously thought, and has brought into focus many previously underrated manifestations, especially kidney tumors.

In 1990 we started an extensive survey of VHL disease cases among a number of French physicians, representative of the various medical specialties involved in the care of people with VHL. This is a continuation and extension of the survey conducted by Dr. François Resche in 1983 among neuro-surgeons. Its ultimate aim is to collect all French cases of VHL disease. In addition, we studied the records of numerous departments of neurosurgery, ophthalmology, pathology, and especially neuro-pathology, dating back in some instances more than sixty years. This proved invaluable for the next step, a formal genetic study involving a thorough, record-based genealogical survey, which enabled us in a number of instances to tie up so-called "sporadic" cases to previously known VHL families.

This project is sponsored by the Association Française contre les Myopathies (the French "Téléthon", to combat muscle diseases) and the Ligue National contre le Cancer (National Cancer Society). The main aim of the project is to promote better knowledge of the natural history, radiology, and pathology, criteria of diagnosis, and management of the health of people with VHL disease.

The multidisciplinary staff is composed of F. Resche (neurology), M. Hurth (neurosurgery), A. Gaudric (ophthalmology), P. F. Plouin (internal medicine), C. Proye (endocrine surgery), J. P. Grunfeld, D. Chauveau (nephrology), Y. Chrétien (urology), P. Hammel (gastroenterology), C. Beigelman, O. Hélénon (radiology), S. Olschwang and G. Thomas (molecular genetics) and S. Richard (clinical genetics). We have contributors to the study in all medical specialties in most French hospitals. Patients are referred to us by physicians who have been alerted by our papers in French professional journals and by the lectures we have been asked to give to various scientific and professional societies.

People with VHL and their at-risk relatives are referred to us at La Salpêtrière Hospital. Basic check-up with neurological examination, ophthalmoscope, and family history is completed along with cerebral and spinal MRI, abdominal ultrasound or CT scan, and 24-hour urine collection for detection of lesions which may not yet have symptoms. Patients are then referred to any required specialist. For the time being, genetic investigation and counseling are done free of charge as part of the research project. Clinical investigation and treatment are paid for by the French state medical insurance system (Sécurité sociale) on an

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80% basis for most people, and on a 100% basis if the patient is deemed "affected by a cancerous disease." Today we know 380 affected people and 85 VHL families in France. Seventeen families have six or more (as many as 26) members with characteristic features of VHL.

Our particular interest is the formal genetics of the disease, that is, the precise way the disease runs in families. The genealogy of each family is studied several centuries back in order to unravel a possible common ancestor who might link various apparently independent families. Large kindreds are a powerful tool for genetic studies, and have led to breakthroughs in other diseases. We have consulted parish and municipal registers of births, marriages, and deaths. Access to recent records for living persons is usually prohibited by law, but permission has been granted to us in view of the scientific and practical importance of this research.

Here is one example of the value of systematic genetic inquiry in daily practice. Patient #654 is a woman, age 27. In 1990 she was operated on for cerebellar hemangioblastoma and treated for retinal hemangioblastomas. Family history was recorded as negative at that time; the patient declined to undergo the investigations that had been suggested by the neurosurgeon. In April 1992 systematic inquiries enabled us to link this patient to a large VHL kindred with 18 known affected members, resulting in fusion of four previously unrelated pedigrees, the "Sancerre" family, originating from a famous vineyard region in the center of France. The father of the patient (#1399) died at age 32, reportedly from malignant melanoma [a cancer not connected with VHL]. He had complained of loss of vision in one eye, dizziness, and headache. Her grandfather (#1398) died in 1970. The relevant entry in the hospital records mentions kidney cancer as the cause of death.

At that point, members of the family gave their consent for systematic investigations. Her brother, age 24 (#2994), had complained for three years of headaches and vomiting, with considerable weight loss, that had been diagnosed at another hospital as a psycho-somatic illness. In May 1992, following the demonstration of a link with a VHL kindred, he underwent tests which revealed a large hemangioblastoma of the medulla oblongata which was successfully removed in July 1992. Check-up of their sister, age 34 (#2993), who had no symptoms, revealed bilateral retinal hemangioblastoma and bilateral renal tumors, that were treated with conservative surgery.

Genetic diagnosis is at present available in France through linked markers when several affected family members have been sampled. It was developed by G. Thomas and S. Olschwang. It is going to transform the outlook for VHL gene carriers, since most of the manifestations of VHL are amenable to effective treatment if diagnosed early. In the future, knowing the characterization of mutations, molecular diagnosis is going to be possible even in the absence of a documented family history.

Drs. Richard and Resche are affiliated with La Salpêtrière Hospital in Paris. Their study is reported in greater detail in a number of professional publications, most recently S. Richard et al, "Renal Lesions and Pheochromocytoma in von Hippel-Lindau Disease," *Advances in Nephrology,* 23 (1994) Mosby-Year Book, Chicago. To assist you in drawing up your own family tree, ask for the booklet *Your Family Health Tree,* available from the VHL Family Alliance. ❑

A *proband,* or *index person,* is the presenting patient. For example, patient #654 gave the family history shown at the top. Genealogical studies fanning out from five individual patients resulted in the determination that all these people were related. Females are represented as circles; males are represented as squares.

Soothe the Body, Heal the Mind

with Jim and Jay Sicard, Florida

At the first meeting of the Massachusetts Chapter at Plum Island, Massachusetts, September 19, forty-five adults and ten children came together to meet each other and share experiences of living with VHL. People came from Massachusetts, New Hampshire, Rhode Island, New York, and Florida.

For some people, it was the first time they had met anyone else with VHL. Even for members of large families, it was usually the first time they had met another VHL patient outside their family. People began sharing stories in the parking lot before they came inside.

There were people there with a variety of experiences of VHL, and with a variety of abilities. One brother and sister, twins age 59, were themselves a good example. They have had the same number of surgeries, in the same places. The brother is doing just fine; the sister has serious walking and speech impairments. Why? We were all eager to learn from one another. What have you been through? How well did it go? What did you learn? If you could do it again, would you have done anything differently? The "pioneers" who have had the most difficulty can provide the richest learning for the rest of us.

Jim Sicard and Mary Lou Egan, massage therapists, gave a talk and demonstration on the use of massage therapy in dealing with the stresses and pains of VHL. It need not necessarily be a professional massage; family members can provide massage therapy to one another. It is a way of communicating your caring, as well as calming, soothing, and relaxing muscles and nerves. As Jim reminded us, the most important ingredient in a therapeutic massage is *intention*. If you approach the person you are massaging with sincere intention to relax them and be helpful, that intention will be communicated through your hands.

The person receiving the massage should lie comfortably on a bed, or well-padded floor, in a place without drafts, where they won't be chilled. The person giving the massage should be working in a comfortable position. Some quiet, relaxing music can add to the experience.

The hands move most easily over the skin if you use some oil. Creams don't work as well because they absorb into the skin or become sticky. A pleasant light oil is best. There are oils made specifically for massage, but even olive oil or corn oil from the kitchen work just fine. Use a small amount, and warm it a little in the oven or in your hands so that it is not cold when it hits the skin.

For people with VHL, there are a few safety precautions. If there are any spinal tumors, you should not put pressure on the spinal column. This is particularly important if there has been any spinal surgery. If there has been any brain surgery, be particularly careful not to put pressure in areas near incisions where the bone may be weakened. And most important, don't do anything which causes pain.

Left to right: Mary Lou, Jay, and Jim preparing Jay for his massage.

In the area of any incision, it is best not to press deeply. Long soothing strokes are nearly always appreciated. Light circular movements to "mobilize" the scar can be helpful in loosening the scar adhesions which may bind skin to muscle and increase tension.

Jim demonstrated a few basic strokes — long gliding strokes *(effleurage)* with flat hands from shoulders to hips, or down the full length of any other muscle; kneading the muscles *(pétrissage)* as you would knead bread dough; or working the muscles in small circles to relax them. It not only relaxes the person, it assists in lymph drainage and helps to rid the muscles of waste products.

Sometimes when muscles are tense or sore, there will be some minor discomfort when the muscles are first kneaded to help them relax. This is normal. If there is any real pain, though, it is time to stop or change to a lighter soothing rubbing stroke.

Altheada J. from New York was timid to try, but agreed to the experiment. She has been managing quite a bit of pain since her second spinal cord surgery in 1989. While she was hoping that it would help, she was worried that it might make the pain worse.

Mary Lou worked closely with Altheada and her husband, Fred, experimenting with different strokes and angles until they determined what was the most soothing and what to avoid. Even though Altheada has no feeling in her legs, there is some deep muscle response, and it proved very soothing to her to have her legs massaged. Mary Lou also touched, just using warm hands and energy. She suggested a massage therapist experienced in Reiki massage.

When Jim first began studying massage and

needed someone to practice on, he found a ready subject in his brother Jay. Jay, age 28, has been dealing with pain since his brain surgery in 1975. When Jay first complained of headaches, his father thought he was looking for a way out of returning to school. "I always hated school," Jay admits. But they proved to be much more than imaginary, and even more serious than the doctor's first diagnosis of migraines.

After the removal of Jay's brain tumor, they thought the road ahead would be clear. But a second brain tumor was found in 1977, and he was diagnosed with VHL. Jay is the first person in his family to have VHL. DNA tests of his parents, brothers, and sister are all negative. In 1984 Jay underwent radiation treatment for control of his spinal tumors. There are more spinal tumors, but doctors are not recommending treatment unless symptoms become more severe. Jay lives year round in Florida, where his parents spend their winters. He and Jim attended the Massachusetts meeting while visiting their parents at their summer home in Massachusetts.

Jim and Jay soon learned that massage was an important ingredient in Jay's pain management. Soothing away the stresses of daily life, and relaxing the tension of the muscles around the pain, significantly reduce his discomfort. They were glad to share what they have learned about massage, and to share other experiences at the Alliance meeting. "The meeting gave us a chance to compare notes, compare histories. It was a chance for comparison, and also for compassion."

Our thanks to Mary Lou Egan of Interlocks Salon, Newburyport, Massachusetts; Jim Sicard, Daytona Beach, Florida; Evelyn and Chuck Sicard, and Tom Salemi, reporter for the *Newburyport Daily News.* ❑

From the Sidelines

Being an unaffected member of a VHL family has been a difficult position at best -- not knowing where or who to turn to as each medical problem arises. For years our family has had to depend on each other's experiences to find knowledgeable, professional medical attention.

Over the past thirty-five years we have had some excellent and, in a few cases, some less than acceptable care. Luckily, we have come in contact with several medical professionals that have worked with us in researching VHL and providing excellent care. I have personally known the frustration of dealing with those that won't believe or even pursue the myriad symptoms that can lead to early intervention in the effects of VHL.

I feel that the VHL Family Alliance and its objectives will be a Godsend to us. I look forward to working with the Alliance. -- *Don M., Mississippi*

Resources

What You Need to Know About VHL, the VHL Handbook, is now available. Copies will be mailed to all members in December. This important booklet describes von Hippel-Lindau disease in simple language, provides hints for managing your health, and a glossary of terms your medical team will use. Developed by a team of VHL family members and health care professionals. Additional copies are available for $1 plus 50 cents postage and handling from the VHL Family Alliance, 171 Clinton Road, Brookline, MA 02146 USA.

Candlelighters Childhood Cancer Foundation maintains a list of organizations that grant wishes for children with chronic or life-threatening illness. Send 50 cents to Candlelighters, 7910 Woodmont Ave, Suite 460, Bethesda, MD 20814.

Starlight Foundation (U.S., U.K., Australia and Canada) brightens the lives of seriously ill children by granting wishes for children 4-18 who have an illness which causes continuing pain and discomfort. There are no income requirements. For more information, contact Starlight Foundation, 75 Saint Alphonsus St., Suite H, Boston, MA 02120, Tel: (617) 739-0020. Canada: (514) 737-4447; England (71) 430-1642; Australia (02) 660-0144.

Brain Tumor Conference set for March 10-13 in San Francisco, March 10-13, 1994, Hyatt Airport. Brain tumor patients, families, friends; and associated health professionals, are invited to attend. Practical information, support, and updates from research. Sponsored by the National Brain Tumor Foundation. For further information, please call 1-800-525-MEET.

For access to the national ***Brain Tumor Support Group Network,*** with more than 100 groups around the country, call the National Brain Tumor Foundation at 1-800-934-CURE. NBTF, 323 Geary Street, Suite 510, San Francisco, CA 94102, (415) 296-0404, Fax: (415) 296-9303.

The ***American Brain Tumor Association*** provides patient services and educational materials, free of charge. Funds research nationwide, and works to educate the public about brain tumors. Publications include a *Primer of Brain Tumors: A Patient's Reference Manual, Coping with a Brain Tumor,* and a newsletter. ABTA, 2720 River Road, Des Plaines, IL 60018, (708) 827-9910, (800) 886-2282. ❑

I remember Mary Margaret Kampmeier, a sweet, laughing young woman with happy dreams for her husband and children. Taken from us suddenly when I was only three by an unknown force which we now know was VHL.

She guards over me and my sisters as we may have to fight our own battles with VHL, and guides us in living a rich and full life until we meet her again. -- *Mary Beth P., Minnesota*

The Many Masks of Adrenal Involvement

by Peggy Graham, R.N.

John and I were married at 18 and 20 — wedding my French-Irish heritage to an athletic Scot. He completed a Ph.D. in chemistry, I finished my nursing degree, and we launched two careers and six children over the next twenty years. We were a healthy lot. One of our few complaints was our constant "battle of the thermostat" — John was very intolerant of heat, and would break out in sweats when I thought the temperature was approaching normal. I attributed it to his chilly upbringing in Scotland, which had left him intolerant of civilized central heating. Beginning about age 40 he was treated for high blood pressure, like so many men his age.

In 1987, John was driving home when he noticed one eye was tearing — but what he wiped from his cheek was blood! He went to an ophthalmologist who sent him to a retina specialist. They told that he had von Hippel-Lindau disease, but that there was no treatment for it. While VHL could also cause brain tumors, there was no need to test for those since "the cure is worse than the disease."[1] They would make themselves known if they needed to be dealt with.

I checked in a few medical textbooks, but the descriptions were not helpful. No one in John's family had any problems with eyes, nor any brain tumors. His mother was healthy, his father retired and died of a heart attack at 67. Perhaps the diagnosis was wrong? In any event, the bleeding episode ended, there was no change in John's vision, and the ophthalmologist didn't give us anything to do, so we promptly put VHL out of our minds and settled back into our happy, hectic daily life.

Two years later, our son Matthew, age 13, began experiencing headaches, restlessness and cold sweats. He would call in a panic from school, declaring himself to be deathly ill. I would go get him, bring him home, and within fifteen minutes he was miraculously cured. I wondered if he were simply "allergic to school" and making up a good excuse to get home. Dramatically, Matthew declared he "was dying," and nobody would believe him. Routine blood work revealed bouncing blood sugars. He was diagnosed as glucose intolerant. His blood pressure and heart rate were normal. But the episodes of panicky calls from school continued.

Meanwhile, John had a heart attack. I was careful to tell the doctors that he had a diagnosis of VHL in the eye, worrying that the blood thinners they were using to dissolve the clot might cause a hemorrhage in the eye. His doctors did not consider that information relevant to his heart condition.

After nine months and no improvement from diet therapy, Matt developed a rapid heart rate. The cardiologist we consulted found a blood pressure of 260/140 — dangerously high! — and Matt was admitted to the hospital. We never suspected that there might be a connection among Matt's blood sugar, his father's heart attack, and the mysterious eye disease mentioned by John's retinal specialist. As part of the comprehensive family history, we mentioned VHL in passing, and the pediatric endocrinologist began to put the puzzle together.

Matt had an abdominal ultrasound and urine tests which revealed pheochromocytomas, or pheos *(fee-ohs),* a kind of tumor on both adrenal glands. Surgeons subsequently removed both his adrenal glands and put him on hormone therapy to balance the cortisone and mineral levels normally regulated by these glands. His condition quickly stabilized and he was out of the woods.

Now that the VHL connection was established, the doctors recommended that the entire family be screened as well — 24-hour urine collections, blood tests, and ultrasounds of the abdomen. John's and most of the children's tests came back normal, but son Jeff (25) was found to have elevated levels of urine catecholamines, adrenal by-products, indicating a malfunction of the adrenal gland.

Jeff had pheos on both adrenal glands, and the doctors felt they had been there for a very long time. He had always been the frail child, but he did not have particularly dramatic symptoms. Looking back, I realize now that he had episodes of symptoms twice before: a period of severe headaches when he was 3, and symptoms which had been diagnosed as hypoglycemia when he was 14-15. This latter problem was attributed to the rapid growth spurt he was going through at the time, and it normalized after several months of watching his diet.

John's own blood and urine tests were normal. For fifteen years he had been treated for high blood pressure, which had seemed to be under control. His heart attack in 1989 was followed by two more in 1990. Clearly something was wrong, but what could it be? If these tests were coming back normal, his adrenal glands must be OK, right? Little by little we began to learn about pheochromocytomas.

Pheochromocytomas are one of the abdominal manifestations of VHL. These tumors of the adrenal gland emit varying amounts of unnecessary adrenalin into the bloodstream in an unpredictable manner. While rarely malignant in the VHL population, they are nonetheless potentially deadly because of the

1. This is certainly not true today. It is very important to get regular checkups for the leading features of VHL, especially eye, adrenal, and kidney, in order to deal with problems before they become critical. See the *VHL Handbook* for details.

effects of excess adrenalin on the cardiovascular system. The symptoms of pheos are variable but may include elevated blood pressure, sweating, anxiety, rapid pulse, facial flushing, frequent urination, and elevated blood sugar levels. With such a variety of effects, pheos are easily misdiagnosed. They mimic symptoms of more common disorders such as essential hypertension (high blood pressure), and glucose intolerance, disruptions in the body's ability to metabolize sugar.

In the past, traditional testing has included abdominal scans, such as ultra-sound, a 24-hour urine collection to test for adrenalin by-products, and blood testing. While abdominal scans discovered many of these tumors, others were missed due to size or location. The tumor does not always produce excess hormones in a steady stream; they may be given off in surges, or "attacks". Unless the blood or urine testing happens to be conducted during an attack, the results can look quite normal.

In the early 1980's a specific test for pheo was developed at the University of Michigan. Called an MIBG, this test involves injection into the bloodstream of a specialized radioactive tracing material (Meta-Iodo-Benzyl-Guanidine) which is absorbed by the pheochromocytoma. This, like other nuclear medicine tests, is a kind of reverse X-ray. Instead of passing radioactive beams through the body onto the film, as in an X-ray, a much lower dosage of radioactive material is injected into the body, to be absorbed by the particular part of the body to be studied. Computerized photographs are then taken which record the location of the radioactive dye. The resulting pictures tend to be much clearer and more precise, and the total exposure to radiation by the patient is much lower, than in traditional X-ray.

On the imaging study, the pheo literally "lights up" and can be readily seen. A few other endocrine tumors can also be identified by this test. A series of pictures are taken over a period of three days, as some tumors are slower than others to absorb the tracer. Since the MIBG tracer material is cleared from the body by the kidneys, patients with impaired kidney function are given smaller doses.

The MIBG test is highly accurate for pheochromocytoma. With MIBG, pheos were finally identified in Jeff, and John, and also in our daughter Lisa, and appropriate treatment plans were worked out for each of them. Unfortunately, John had already sustained permanent heart damage, but our children's conditions were identified early enough to avoid permanent damage.

In 1986, during an ultrasound check for cysts on her ovaries, our daughter Lisa was found to have a tumor on her right adrenal gland. Her urine and blood tests were normal, however, so it was declared a cyst or benign tumor, and was left alone. Following this new MIBG testing, Lisa had one pheo removed.

Episodes of sweating, "hot flashes" or inability to tolerate heat, facial flushing, and the racing pulse that Matthew experienced are typical symptoms of pheo. However, they may not always be present. John and Jeff both seem to have adapted to the pheo surges. Instead of characterizing the surge as a "hot flash," John was more generally intolerant of heat.

In the largest study of pheochromocytomas every undertaken,[2] Dr. Neumann and his team at the University of Freiburg demonstrated that traditional blood and urine testing for pheos found only about 40% of the tumors. CT was more useful (72%), but the most reliable tests were MIBG and MRI (95%). While these tests are more expensive, they are so much more accurate that the additional expense is well worth the potential saving in cardiovascular damage.

Moreover, the Neumann study showed that among people with seemingly random pheochromocytoma, 20% proved to have VHL — four times the number they would have expected. Conversely, of the 79 VHL subjects studied who did not have symptoms of pheos, 46% proved to have pheos, nearly four times the expected rate. There are familial variations -- some families tend to have more pheos than others. But everyone with VHL should be screened for pheo, especially before any kind of surgery. The stresses of surgery can cause a pheo attack and complications.

We are hopeful that increased awareness of the more accurate tests documented in the Neumann study will lead to earlier and more accurate diagnosis of pheochromocytoma, and of VHL.

Both Matt and Jeff have had both adrenal glands removed and are on replacement therapy. Matt's body handles it quite well, but Jeff's cortisone level is very unstable. Whenever he has a cold, or is under a lot of stress, or on some medication, he must increase his cortisone dosage to avoid trouble.

For this reason, Dr. Neumann and Dr. John Libertino[3] at the Lahey Clinic, Burlington, Massachusetts, now recommend whenever possible removing the pheo only, and leaving as much remaining adrenal tissue as possible, to minimize the need for replacement therapy.

Now that we know that we have VHL to deal with in our own family, and now that we understand its many features, we all go regularly for checkups and work actively with our medical team to determine the action plan for dealing with issues as they arise.

2. Hartmut P. H. Neumann et al, "Pheochromocytomas, Multiple Endocrine Neoplasia Type 2, and von Hippel-Lindau Disease," *New England Journal of Medicine,* 329:21, Nov. 18, 1993, pp. 1531-1538.

3. Malone, Libertino, et al, "Preoperative and Surgical Management of Pheochromocytoma," pp. 567-582 in *Adrenal Surgery,* ed. Libertino and Novick, *The Urologic Clinics of North America,* 16:3 (1989).

Our thanks to Dr. Brahm Shapiro, Dept of Nuclear Medicine, Univ. of Michigan, for his assistance in the preparation of this article. ❑

Health Care Reform, U.S.A.

As we work with families across the United States it is clear that the current system of health care funding is not working for many, if not most, families with VHL. While most of us are agreed that change is needed, it is important to look at the proposals and envision whether our own health care situation would be better or worse under the new plan.

There are a number of organizations lobbying in Washington for special interests. We went looking for organizations which would represent the needs of people like ourselves, to keep us informed, and through which our voices could best be heard. We report here on two such organizations.

Families U.S.A.

1334 G Street, NW, Washington, DC 20004, Tel: (202) 737-6340, (800) 593-5041 from most states.

Families U.S.A. is a well-funded lobbying group, focused on passing President Clinton's plan. Their position is that while the plan may not be perfect in every aspect, it is such a very good first step that it deserves to be passed now, and fine-tuned later.

Phyllis Torda of Families U.S.A. told us that they will be monitoring amendments to the bill as it passes through Congress, working to ensure the maintenance of its essential characteristics:

- Does it guarantee that you and your family won't ever lose your health insurance, no matter what?

- Does it include help for your family to provide long term care to a father with Alzheimer's disease or a mother with crippling arthritis at home where you want them cared for?

- Does it do something about getting skyrocketing health charges under control?

- Does it give you somebody to complain to if you're not happy with the service you get from your doctor or hospital?

Family Voices

P.O. Box 769, Algodones, NM 87001 Tel. 505-867-3159, Fax: 505-867-6517.

Family Voices is a grass-roots organization composed of families who have children with special health needs, and their friends. While most lobbying groups focus on routine health and accident, and the needs of the elderly, this group focuses on the needs of families like ours which will require access to preventive diagnostic tests, extended therapy, or health services due to chronic illness for people younger than retirement age.

They distribute accurate information about proposed changes in health care. Polly Arango of Family Voices told us that "while our bottom line is that health care reform must take place, and universal coverage and the abolishment of pre-existing conditions clauses is wonderful, it is clear that our children and families are still extremely vulnerable, and in many ways remain forgotten. That has got to change! There is still work to be done."

They ask that every family read the September 7 draft of the Act, and then sit down and write their own analysis of how they and their family would fare, comparing health services and costs in the President's plan to what they have now. So far, more than fifty families have responded to this appeal.

What Can You Do?

Read the September 7 draft of the proposal (250 pages), which is much more readable than the legislative version (1600 pages). You will find it in most book stores as *The President's Health Security Plan* (Times Books, $8). In reading the proposal, see how well it fits the needs of your own family. A few changes have been made since the September 7 draft, which are being reported in the press.

As you read it, consider how your family will fare. Send your comments to Family Voices, and/or Families U.S.A. Please send us a copy of your letter. Both organizations maintain databases of family stories which can be used in Congressional hearings as examples of why a particular provision is needed.

Visit and write your members of Congress. They all come back to their home districts frequently. Either of these organizations can provide a list of Members of Congress, or call the Congressional Switchboard at (202) 224-3121. Most Senators and Representatives will hold health care reform meetings at home, so get on their agenda. Tell your own story, and ask questions.

Talk to friends, neighbors, family members, and your family's health care providers about what's going to happen with your family under the plans.

Be an active participant in this important legislative process. ❑

Family Leave Law Now in Effect

The Family and Medical Leave Act (U.S.) went into effect in August 1993. The act gives workers the right to take an unpaid leave of absence without jeopardizing their jobs. Private employers with fifty or more employees are covered under the law.

You can qualify for family or medical leave when a son or daughter is born or adopted; when there is a need to care for a seriously ill spouse, son, daughter, or parent; or when you have a serious health condition yourself. Although the leave is unpaid, your employer is required to continue paying your health insurance premiums at the same rate while you are out. *A Guide to Using the Family and Medical Leave Act: Questions and Answers,* is available for $4.95 from Women's Legal Defense Fund, 1875 Connecticut Avenue, N.W., Suite 710, Washington, D.C. 20009. ❑

Introducing the Board of Directors

Lois Erickson, Bloomington, Minnesota

I am married and have two children: Carmen and Chad. I have VHL and so does my son Chad. I work at Knutson Mortage Corporation in the Master Servicing Department. I have many hobbies and think of myself as a healthy person. My story is in the June issue.

I am delighted to be a member of the VHL Family Alliance, and think the organization has given me a purpose in life. I don't want anyone to be as uninformed as our family was for 40 years. I will do my best to talk about VHL to everyone who will listen. I am thankful for each day that I feel well and can continue going 100 miles per hour!

With Cindy Dearing of Missouri I will be chairing our Annual Meeting in Kansas next spring. I look forward to meeting all of you there!

Joyce Graff, Brookline, Massachusetts

I work for Digital Equipment Corporation, as a software product manager with Electronic Mail. I am a past Director of the National Braille Association and the Massachusetts Association for the Blind. I am a member of several community groups and industry organizations in telecommunications.

I signed on for life with VHL in 1961, when I became engaged to Frank. Frank's outstanding academic career and success as a history professor in the face of the challenges VHL and blindness threw his way, were inspiring to those of us who were closest to him. It was his wish that we learn as much as possible from his experiences so that the next generation would stay healthier.

I am delighted to work with the Alliance to make the road easier for our son, and for all of us who live with VHL. My son and I are enormously grateful to our many new friends in our "other family," and to the many physicians who give so generously of their time to help us with the newsletter, handbook, and other materials.

Peggy Graham, Warren, Michigan

I am a nurse in maternal and child health at the Bi-County Community Hospital in Warren, Michigan. I am a lactation consultant and coordinate teaching programs for new mothers, and the nurses who work with them. I am also a busy mother and grandmother.

My family has had a crash course in VHL, beginning only five years ago. Since then, in addition to adrenal involvement, we have had brain and spinal aspects as well. Our adrenal story is in this issue. It is difficult managing the health care of so many people all at the same time, but somehow we manage it all! We are delighted to have found so many other wonderful families. Together, we will overcome VHL.

Hazel Murfitt, Ely, Cambridgeshire, England

I am a wife and mother of people with VHL, with more than twenty years of brain, spinal, kidney, and pancreas involvement. Our story is in the September issue.

Since early in 1993, when I decided to start a contact group for VHL patients and their relatives, I have taken a number of courses to improve my skills in helping others with this challenging disease. Since then we have been joined by an established group previously coordinated by Mr. Victor M. in Wales. While we do not have group meetings here, I am available by telephone 24-hours a day for VHL families who need to talk.

Our family is very active in spreading the word about VHL in the press in England. Ken and I were honored to open the Royal Flower Show at Hampton Court in June.

Susan Warnick, Reisterstown, Maryland

I have been dealing with VHL since my husband Craig's diagnosis 19 years ago. Engaged at the time, VHL totally changed both our lives. Shortly after Craig's diagnosis I attended nursing school and began working in pediatric neurology and neurosurgery at Johns Hopkins Hospital in Baltimore. I continue to work with children in neurologic rehabilitation at Kennedy Krieger Institute.

Over the years I became aware of the problems faced by VHL families. I was committed to establishing an organization for VHL patients and their families, but never had the time with the frequent surgeries in our own family. I have been delighted to join forces with other VHL families with the same dream and form the VHL Family Alliance. Craig and I are featured in Dr. Ben Carson's autobiography, *Gifted Hands.* A piece of our story is in the September issue. We have one child, Chloe, who has brought us tremendous joy and renewed strength in our battle with VHL.

Darlene Young, Plainville, Massachusetts

I am the Administrative Assistant at the Council on Aging in Attleboro, Massachusetts. I am also an elected member of the Housing Authority of the Town of Plainville, where I serve as Treasurer.

My husband, Bill, and one of our two sons have VHL, as do a number of Bill's relatives. We have had brain, eye, kidney, adrenal, and spinal involvement to deal with. Our family has learned over the years to support one another in managing this challenging disease. We are delighted to be able to work with the VHL Family Alliance, sharing what we know, and learning from all of you. ❑

Alliance News

1993 is quickly coming to a close, and our first year of operation is nearly done. Be sure to send your membership by the end of January 1994 to be a ***Founding Member!*** We will still welcome new members, *of course!* but those who helped to pioneer this organization during this start-up year will always be special to us.

Kansas/Missouri Chapter

The Kansas/Missouri Chapter met in October in Kansas City, with 21 people present, sharing their stories and their questions. There are now state contact persons in 17 states. Call our 800 number to reach the coordinator in your area, or to volunteer to be a state contact or coordinator.

The Year in Review

Our primary goals this year were:

- to reach our and find VHL families throughout the country, and begin to pool our learning;
- to make contact with VHL families in other countries, to learn too from other cultures, other health care systems;
- to establish a base of educational materials;
- to raise the level of awareness of von Hippel-Lindau disease among medical professionals and the general public.

We feel we have made a very good start, and look forward to building upon these beginnings in 1994.

Reaching families in North America

We are now in contact with nearly 1000 affected people in the United States and Canada. A large percentage of these are members; others are reached through physicians and genetics counselors.

Contacting other countries

Our sister organization in England is in contact with numerous families in England, Scotland, Ireland, Wales, and Canada.

We are now in correspondence with physicians in some twenty countries, and are beginning to reach families in those countries. As shown in the article from France in this issue, we are interested in learning the experiences of VHL in many countries, the familial variations which may be clearer there than stirred together in America's "melting pot," and the differences in treatment among the different health care systems and different medical traditions.

Publications

We met our publishing goals of: four issues of the *VHL Family Forum;* two pamphlets: *Your Family Health Tree* and the *VHL Handbook*; two brochures: *What is VHL?* and *Not So Rare After All.*

We now have good, portable, easy to read answers to questions like "What is VHL?"

Awareness

We made a good beginning. More than 20 telephone and computer information services now know about VHL. We displayed brochures at medical meetings, and were listed in numerous professional publications, which brought inquiries from a wide range of social workers, genetic counselors, nurses, and medical professionals. Several members were successful in getting their stories published in local newspapers.

The mainstream press has been slow to pick up news about VHL -- news which is so very interesting to us but which affects a small percentage of TV viewers. Nonetheless, a great many more medical editors now recognize the name "VHL" than this time last year, and that is a valuable first step.

At their November meeting, we were accepted as a full member in the National Organization for Rare Disorders (NORD)!

Pat Yourself on the Back!

Thanks to you -- your help, your funding, your encouragement, we are off to a wonderfully strong beginning. We look forward to your continued participation in the coming year -- it is your willingness to share your learning -- your experiences or your professional research -- which are helping to bring together a wealth of information about VHL which has never before been available. We give extra special thanks to our many friends and unaffected relations whose continuing financial and moral support make this work possible.

Join us for the First Annual Meeting!

We look forward to our Annual Meeting in Kansas City the weekend of April 16-17. Individuals and families affected with VHL, their friends, and health care professionals from several countries will have an opportunity to share their experiences, ideas and questions about VHL. Leading researchers will share the latest developments in diagnosis and treatment.

Most important, it will be an opportunity to get practical information and support from others who are dealing with this challenging disease. It won't be the same without ***you!*** The specific session announcements and information for registration will be distributed in February.

Goals for Next Year

According to the geneticists, there are 6000 people with VHL in the United States alone. We want to find the other 5000 people, make sure they have a diagnosis of VHL, and the information they need to maintain their health.

With your help, we will be undertaking publicity and projects to help make VHL a household word. The more we can do to make people think of VHL as a possible diagnosis, the more likely we are to find the other people who are out there dealing with a frustrating collection of seemingly unrelated symptoms, with no weapons to combat this disease.

Pick a project -- we need your help!

Thank You!

Many thanks to those who helped with programs and with the current issue

In Honor of . . .

...Damon Graff's graduation from Brown University, from John, Eileen, and John Kellogg Hodgman
...The 50th Wedding Anniversary of Martha & Oliver Jeanes, from June & Harry Wilcox

In Memory of . . .

...Richard A. Cohn, M.D., from Sidney & Dorothy Cohn
...John E. Garofalo, from his wife Catherine
...Dr. Frank W. Graff, from Ron Nash & Judy Stacier
...Dr. John C. Graham, from his family
...Mr. & Mrs. Lonzo E. Hayes, Sr., from Mr. & Mrs. Sheldon C. Marshall
...Harry Horelly, from Rachael & Joyce
...Mary Margaret Kampmeier, from her daughters, Mary Beth Paulson, Laurie Dripps, and Susan Wanger
...Jean & Dr. Williams Leebens, from June & Harry Wilcox
...Zinaida "Bula" Simonovsky, from Joyce & Rachael

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United Network for Organ Sharing (UNOS)
Betsy Wilson, Let's Face It!, Concord, Massachusetts
... and Members of the VHL Family Alliance

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May the Gifts of Peace, Joy & Hope be Yours this Holiday Season!

We have enclosed your holiday gift -- your own new VHL Handbook.

Please consider sending your gift to help us continue this work.

The VHL Family Alliance is dedicated to improving diagnosis, treatment, and quality of life for people with VHL. Please support our vital programs: information, hotline support, education, advocacy. Ask friends and family members to give donations instead of ties and handkerchiefs.

We are all volunteers. We have no paid staff. 100% of your donation goes directly to provide services. All contributions are tax deductible in accordance with IRS regulations.

This is ***your*** Alliance. We need ***your*** help. ***Thank you!***

Enclosed is my gift to support the programs of the VHL Family Alliance.

- ❑ $1000 pays for one issue of the newsletter
- ❑ $500 pays for toll-free hotline support for two months
- ❑ $100 puts VHL brochures into the hands of doctors, nurses, patients
- ❑ $50 sends 10 information packets to libraries and genetic counselors
- ❑ $25 pays for mailings to one member for a year

Please make checks payable to VHL Family Alliance

Name: _____

Address: _____

_____

City: _____ State: _____ Zip: _____

Country: _____ Fax: _____

Phone (home): _____ Phone (work): _____

Thank you!

. All Members receive a membership certificate, one copy of each publication.
. Free subscriptions are available where the dues are a hardship.
. Donations greater than $250 receive a special sponsor certificate.
. Memberships received by end of January 1994 are Founding memberships.

- ❑ My employer will match my donation. I have enclosed the necessary form.
- ❑ This is a NEW ADDRESS - please change your records
- ❑ Audio version needed for a handicapped reader
- ❑ My occupation is _____

I am a
- ❑ VHL patient
- ❑ VHL family member
- ❑ Supporting Friend
- ❑ Sponsor (donation greater than $250)
- ❑ Professional (please specify)
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- ❑ I am interested in participating in a local support group

My contribution to the VHL Family Alliance is given

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Please send an acknowledgment card to:

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In Britain, send to VHL Patient and Relative Contact Group, 114 Longfield Rd, Littleport, Ely, Cambs CB6 1LB

There is no place like home! Dorothy and Toto, accompanied by the Cowardly Lion, the Scarecrow and the Tin Man, sought help from the Wizard of Oz to get Dorothy back home to Kansas, in Frank Baum's classic book. Illustration © 1990 Top Art, Inc. Derby, Kansas. From
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Join us in Kansas City, April 16-17, for the First International Patient/Provider Conference on VHL!

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