

## Down's Syndrome May Hold Important Clue

with excerpts from a talk by Judah Folkman, M.D., Children's Hospital, Boston, at the American Urological Association, 98<sup>th</sup> Annual Meeting

The study of the basic processes behind new blood vessel growth began in earnest during the late sixties and has since produced a sizable body of knowledge. Today, many pharmaceutical companies are producing drugs that can effect change in both primary cancer sites as well as the spread of metastatic disease. Angiogenesis is the creation of new blood vessels. Angiogenesis is a normal and necessary function of the body for wound healing, and during menstruation and pregnancy. In VHL and in cancer, that normal function can go out of control in certain cells, and knots of blood vessels are created where they are not needed, forming hemangiomas or feeding cancer tumors.

In presenting this year's John Duckett Memorial Lecture at the American Urological Association, Judah Folkman, M.D., said that the field of angiogenesis research has grown considerably since 1971 when there were only three papers in the literature with angiogenesis in the title, two from his lab and one criticizing a former paper of his.

"This has expanded to thousands of laboratories worldwide, and there are on average forty papers every six days published in the scientific literature that have angiogenesis in the title," said Dr. Folkman, Professor of Pediatric Surgery at the Children's Hospital and Harvard Medical School in Boston, Massachusetts.

The role of positive and negative regulators in angiogenesis is becoming clear and the study of oncogenes (cancer-promoting genes) has produced surprising results. The mechanism of these genes can be quite complicated. "In fact, many of the oncogenes up-regulate a stimulator and also down-regulate an inhibitor. It has been shown that at least twenty percent of the known oncogenes are essential for the angiogenic switch," said Dr. Folkman. The "angiogenic switch" is his term for the moment at which the tumor begins to recruit new blood vessels so that it can grow. A tumor needs a food supply in order to grow, much as an army travels on its stom-

ach. Without a supply-chain of blood vessels to maintain the food supply, the tumor will stop growing at a very small size.

The original discovery of oncogenes and tumor suppressor genes was based mainly on their ability to promote or decrease tumor growth in laboratory dishes. A recently discovered new function of oncogenes and tumor suppressor genes is that they also regulate angiogenesis. VHL is a tumor-suppressor gene whose job is to be part of the "off" switch for angiogenesis. When the VHL protein is missing in the cell, the "off" switch doesn't work and the process of angiogenesis begins.

In experimental animals, microscopic tumors must grow, or they cannot spread. The offspring of this research is now producing treatments for cancer in the human body by either directly or indirectly interfering with various angiogenic switches. The Nova Series of WGBH Boston, an affiliate of U.S. National Public Television, has prepared a fascinating report on angiogenesis, called *Cancer Warrior*, describing Dr. Folkman's development of the theory of angiogenesis. The story is also recounted in the book *Dr. Folkman's War*.

One of the most studied inhibitors, endostatin, continues to offer surprises. With the relative ease that laboratories now have in producing endostatin (some 81 have), the rate of publication is increasing by three or four per week. Endostatin is much in the

cont'd on page 2

### Inside this issue!

Boston Meeting Nov 2003	Genetics Home Reference
Grandmother	Support and Awareness
Lifestyle Changes	Ask the Experts: Eye Issues
Early Eye Testing	Uninsured at Disadvantage
CCC in New York	Local Activities
CCC in Houston	Ask the Family
Beta-Carotene & Smoking	VHL Tissue Bank
Resources	2004 Symposium in Japan

news, and sounds very exciting indeed. However the connection between endostatin and VHL has not yet been made. In 1994, Joyce Graff and Dr. Lloyd M. Aiello of our Medical Advisory Board met with Dr. Folkman and asked whether endostatin might be helpful for people with VHL. Dr. Folkman indicated that there were tracers in the urine which should tend to indicate whether endostatin might be beneficial.

VHLFA Members donated some 28 urine samples to do the assessment. Dr. Frederick Hes of Leiden in the Netherlands, then a fellow in Dr. Folkman's lab, examined these samples and determined that endostatin was not likely to be helpful for people with VHL. The urine test assessment is described in the Nova program, where researchers examined mouse urine to find endostatin. In subsequent research, however there is no proof that this urine test is definitive for humans. Angiogenesis is indeed the mechanism in which VHL participates, and with the complexity of this mechanism, endostatin cannot yet be completely ruled out.

An intriguing insight has been gained by studying the apparent low cancer rate in patients with Down's syndrome. "A recent study of 17,000 age-matched American Down's patients showed that they have virtually no prostate cancer, virtually no breast cancer, virtually no pancreatic cancer, a mild leukemia, and the same incidence of testicular cancer." Dr. Folkman asked: "What does that mean in the big picture analysis?"

As it turns out, Down's patients have an extra copy of collagen 18 on chromosome 21 and endostatin is an integral component of collagen 18. The amount of circulating endostatin in these patients' blood is ten times greater than in the normal population. And the effect is felt on very different tissues and cancer types. Would the extra endostatin in the blood of a person with Down's syndrome effectively control VHL?

Is there a VHL family with a member with Down's Syndrome? This person might seem to be unaffected by VHL. DNA testing might be able to tell us, however, whether this person in fact has the VHL alteration, but that it is controlled by the extra endostatin created by Down's Syndrome. Please write to Joyce Graff, [editor@vhl.org](mailto:editor@vhl.org), if you have a family member willing to get DNA testing for VHL and share their results with all of us. Similarly, if a person with Down's does get VHL tumors, then it would tend to rule out the effectiveness of endostatin with VHL. Whatever the results, these answers would significantly shorten a clinical trial.

In the cells of someone with VHL, there is one altered copy of the VHL gene which is not making the correct VHL protein, and one copy that does make sufficient VHL protein to keep cell growth

regulated correctly. If something happens to knock out this second healthy copy of the VHL gene, then a process of angiogenesis begins. In most cases, the body's own self-repair mechanisms take care of the problem. If not, however, where there is too little VHL protein, then there is too much Vascular Endothelial Growth Factor (VEGF). Much drug development work has focused not directly on VHL, but on VEGF. If we can control the amount of VEGF in the cell, we can also control cell growth. There are several new VEGF inhibitors now in clinical trials that may be helpful with VHL. Examples include PTK787 (Novartis), SU11248 (Sugen), and Avastin (Genentech).

"Angiogenesis research is providing a unifying principle for diseases which, although they have different names and are being treated by different specialists, are dominated by the same pathologic process – uncontrolled angiogenesis." We have long wondered what connected the many tumor types involved in VHL. Dr. Folkman concluded with this analogy: "If you should happen to drain the Pacific Ocean, you should not be surprised to find that the islands are connected." Angiogenesis is the connection among VHL tumors and other cancers.

The Nova program is available on the internet as video or plain text at [www.pbs.org/wgbh/nova/cancer/folkman.html](http://www.pbs.org/wgbh/nova/cancer/folkman.html)

The program is also available on DVD from our bookstore, [www.vhl.org/bookstore](http://www.vhl.org/bookstore)

See also the book *Dr. Folkman's War*, by Robert Cooke, Random House, 2001, [www.vhl.org/bookstore](http://www.vhl.org/bookstore)

## Boston meeting

November 1, 2003, 9-5

Auditorium, Lahey Clinic

Presentations will feature physicians and researchers from the three Clinical Care Centers in Boston: Brigham & Women's, Lahey Clinic, Massachusetts General Hospital, and including Children's Hospital, Joslin Diabetes Center, and the Dana-Farber Cancer Research Institute.

The Program will include news on angiogenesis inhibitors, kidney treatments, brain and spinal tumors, living with VHL, and lots of time to talk with other attendees.

Hotel rooms are being held at the Burlington Marriott, 1-800-872-4683 or 1-781-229-6565. Call to make your own reservation. The VHL conference rate is \$79 per night for up to four people in one room.

Conference registration will be \$25 per person, which includes lunch and breaks

CEU credits will be available for genetic counselors and nurses.

# On Becoming a Grandmother

by Lee Ann K., Washington

What a blessing and a joy to be present as my first grandchild Savannah came into the world on May 8, 2003! She was so tiny and so very beautiful. As I looked into that little face and watched the joy on my son's face as he saw her for the first time, I wanted to sing and dance. I was a Nana! I was of course very circumspect on the outside though so as not to get tossed out on my ear by a very tired and emotional new mother.

I felt so many emotions that day. I believe the most prominent one was of gratitude. I was so grateful that I had reached the age of 46 and was alive to see this miracle. I felt grateful to a Heavenly Father who has given me this treasure and helped me find the joy and the blessings in my life with VHL. The gratitude that I feel every day for the doctors, researchers, and all the medical staff I've met over the years is very hard to express. Because of their devotion and skill I am still here -- Several surgeries under my belt, but not too much the worse for wear!

My father had VHL, but was not diagnosed during his lifetime and died at the age of 30 when I was 3 years old. His mother, my grandmother, was blind and had a brain tumor, but was never diagnosed with VHL during her lifetime. She died when my father was a little boy. Neither of them lived to see their children grow up, let alone their grandchildren.

My mother asked the doctors when I was growing up if I would also get a brain tumor. She was told it was just a fluke of nature that my father and grandmother both had them. I feel so much pain for my dad and my grandmother at what they must have had to go through, and pain for myself when I think of never getting to know them.

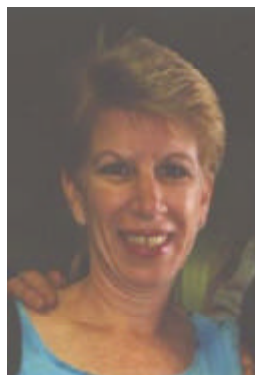
In 1993 I had my first brain surgery and in 1994 after having 5 kidney tumors removed I was finally diagnosed with VHL. At this point it was a blessing to find out about the VHL Family Alliance. Their support really helped me to learn and deal with this disease. In 1997 I had laser surgery to remove a tumor in my eye. Then we received a huge blessing. I was invited to join a protocol at the U.S. National Institutes of Health (NIH) in Bethesda, Maryland. In 2001 Radio Frequency Ablation (RFA)<sup>1</sup> was performed on one of my kidneys to remove another tumor. It was so much more non-invasive and I was only in the hospital for one and a half days. Following another brain surgery in 2002 to remove Buford, Beulah, and Buster (I always name my tumors) I was invited to join another protocol. I am so grateful to

all the folks at the NIH. They have become like a second family to me.

Today I am doing wonderfully well! We are only watching 1 new little brain tumor (Eula May) but she is very tiny and we just aren't giving her any fertilizer! I feel great! Last June I was able to quit my job to help with my husband's business, paint, write, and look after an 11 year old niece we're raising. I have launched a small business in motivational speaking. It's been a great year and I feel better physically than I have in a long time. I no longer have the stressful job or a two hour a day commute. I've been very fortunate to have full recoveries after each surgery. I think I'm pretty normal, but who knows what my family would say!!

When I compare my life today versus the lives of my Grandmother and my father, I feel like a pioneer. I haven't trekked the Oregon Trail in a covered wagon but I have been on a journey that my VHL-affected father and those before him were never able to take. I have been blessed enough to watch my children grow up and now I'm blessed to share the lives of my grandchildren. I find great joy in my life and I believe it really couldn't get much better than this. I am so thankful to all the people who strive to make this possible for a new generation of pioneers.

1. Radio Frequency Ablation (RFA) is the process of using a laparoscopic heat probe to "cook" kidney tumors in place. See VHLFF, Sept 2000.



## We Need Your Help!

VHLFA is supported almost entirely (95%) by individual contributions from people just like you. Please help us any way you can.

- \* Combined Federal Campaign (CFC #1098)
- \* Write in VHLFA in your United Way drive as "donor choice"
- \* Donations in honor of a birthday or anniversary.
- \* Donations in memory of a loved one.
- \* Gifts of appreciated stock
- \* Remember VHLFA in your will.
- \* Help plan or run a fundraising event.

Thank you!

## Lifestyle Changes can halt cancer

A new study presented during the Moderated Posted Sessions at the American Urological Association meeting in Chicago showed that lifestyle changes, including regular exercise and a low-fat diet, may slow, stop, or even reverse the progression of prostate cancer. We do not yet know what implications this may have for other cancers.

The study represents the first randomized, controlled clinical trial of the effect of lifestyle changes on the progression of prostate cancer, as measured by prostate-specific antigen (PSA)<sup>1</sup> levels. Ninety men with biopsy-documented early prostate cancer who chose watchful waiting over conventional treatment for reasons unrelated to the study were randomized into either an intervention group or a control group.

"The patients in the intervention group were able to make comprehensive lifestyle changes for at least one year, and the results were consistent that this intervention may have stopped or perhaps reversed the progression of prostate cancer," said Dean Ornish, M.D., President and Director of the Preventive Medicine Research Institute of the University of California, San Francisco (UCSF), who presented the study results.

The intervention included exercising (mostly walking) at least three hours a week for 30 minutes per session; a strict low-fat vegetarian diet emphasizing unprocessed whole foods; and stress-management techniques that consisted of meditation, gentle stretching, breathing techniques, guided imagery, and progressive relaxation.

"After a year, PSA decreased 4 percent in the experimental group and rose 6 percent in the control group," Dr. Ornish said. The top one-third of the most compliant patients had 13% better PSA scores than the lowest third.

Adherence to the lifestyle interventions also correlated with inhibition of Lymph Node Carcinoma of the Prostate (LNCaP) cell growth across both groups by a factor of 7 to 1," he said. LNCaP cell growth was inhibited in 70 percent of patients in the experimental group versus 9 percent in the control group.

The UCSF researchers plan to continue to follow all the patients in their study to determine how many undergo conventional treatment and the rates of metastasis and survival.

1. The PSA is a blood test that indicates the level of activity or danger in prostate cancer. This is called a "marker", an indicator in blood or urine that provides a clue as to the activity inside the body.

## Early Eye Testing is Important!

by Eileen M., Brisbane, Queensland, Australia

When I was 3 we lived in New Zealand. I used to walk into things, and my family thought I was just not watching where I was going. At my three year nurse check, they did an eye test. When the nurse covered my left eye, I panicked and said I couldn't see.

My parents took me to a hospital in Wellington where they said I had retinal angiomas. The specialists there did not know what to do and sent me home.

A few months later there was a program on the TV about Retinal Angiomas and a Professor from Christchurch Hospital was being interviewed. It was a Saturday night and the next day my mother and I were on a plane and at the Eye Dept first thing Monday am. Well then things started moving as the Professor had just come back from overseas and did all these tests on me and after being there for a few days we were told that I had VHL.

There were many trips to that Hospital and I had cryotherapy to stop the angiomas bleeding and vitrectomy to clean out the blood. The whole family was tested and they did not find VHL in my parents or sisters. If only they had found the VHL earlier in my eyes, they might have been able to save my sight in that eye.

In addition to VHL, I have a second condition, juvenile osteoporosis, which has caused me at least as many problems as the VHL. You would think VHL would be enough, but I guess I won the genetic lottery! Despite all this I have a great life! I'm currently a third year student at the University of Queensland doing a Bachelor of Applied Science in Animal Studies. I have wonderful, beautiful, fun, inspiring friends, a loving, supportive family, and a truly amazing boyfriend, who all understand that sometimes I'm not at my healthiest but they all help me out whenever I need them to – whether I ask or not. I have a great love of music, and really enjoy going to live shows. I love being with, and learning about, animals. I strive to have as much fun as possible at all times, and I am also keenly interested in natural therapies, auras and the supernatural. I consider myself a positive, friendly, kind person. Hopefully I can obtain a lot more inspiring lessons out of what life hands me and share my experiences with others. I hope you take something positive away from this site.

Eileen M.  
Brisbane  
(right)

## New Clinical Care Center in New York

By Wendy Chung M.D., Ph.D., Columbia University, New York, NY

A new VHL Clinical Center has recently opened at Columbia University and the New York Presbyterian Hospital in New York City. The comprehensive clinical center will provide coordinated care for all aspects of a patient's clinical care including diagnostic imaging and surgical intervention.

Initial contact with the medical center and care coordination is provided through the oncogenetics office, directed by Dr. Wendy Chung. Donna Russo, a cancer genetic counselor, speaks with each patient on the phone to answer questions and determine the patient's individual needs. If genetic testing is required to confirm the diagnosis of VHL or to test pre-symptomatic family members, Dr. Chung facilitates the counseling and genetic testing.

Patients come to the VHL Clinical Center and are seen by multiple specialists and have tests done on the same day. Multiple members of the same family can also be seen on the same day. The most advanced imaging methods are available along with a full team of ophthalmologists, urologists, general surgeons, neurosurgeons, and endocrinologists to facilitate surveillance and management of all manifestations of VHL. The VHL team meets to coordinate procedures and follow up care of the patient, and the patient receives a written summary of the recommendations of the team. Our group of specialists works together to offer exceptional patient centered clinical services.

The VHL center can be reached by email at [wkc15@columbia.edu](mailto:wkc15@columbia.edu) or by phone at +1 (212) 305-0190.



## New Clinical Care Center in Houston

By Eric Jonasch, M.D., MD Anderson Cancer Center, Houston, Texas

A new VHL Clinical Center has recently opened at the MD Anderson Cancer Center in Houston, Texas. The Clinical Center aims to draw on the MD Anderson Cancer Center's breadth of experience in managing all aspects of patients' needs, using a comprehensive roster of specialists possessing skills specific to patients and families living with VHL.

Initial contact with the Clinical Center occurs through the Genetics Counseling department, led by Dr. Louise Strong, a leader in Cancer Genetics. Four Genetics counselors work in the department, including Molly Daniels, the contact person for patients making inquiries to the Clinical Center. Coordination of care occurs in association with the Genitourinary Medical Oncology department through the office of Dr. Eric Jonasch, a clinical researcher with a specific interest in renal cell carcinoma.

A schedule tailored to the specific needs of patients and families will be generated. The team of physicians associated with the MD Anderson VHL Clinical Center will be contacted to generate screening, treatment and follow up visits necessary for short-term and long-term care.

A number of specific techniques and skills uniquely suited for patients living with VHL exist at the MD Anderson Cancer. These include minimally invasive surgery, performed by Dr. Surena Martin in the Urology department. Dr. Kamran Ahrar has extensive experience in radiofrequency ablation.<sup>1</sup> Dr. Franco DeMonte is an expert neurosurgeon with experience in the management of the neurological manifestations of VHL. Dr. Douglas Evans is a leader in pancreatic surgery, and has long dealt with the pancreatic manifestations of VHL. Dr. Morris Groves represents the Neuro-Oncology department, and Dr. Bitu Esmali brings her experience in Ophthalmology to the VHL Clinical Center.

The MD Anderson Cancer Center has state-of-the-art imaging and therapeutic facilities. Together with the team of physicians and counselors in the VHL Clinical Center, the goal is to deliver state-of-the-art therapy in a caring environment. The VHL Clinical Center can be reached by email at [cgc@mdanderson.org](mailto:cgc@mdanderson.org) or by phone at +1 (713) 745-7391.

1. Radio Frequency Ablation (RFA) is the process of using a laparoscopic heat probe to "cook" kidney tumors in place. See *VHLFF*, Sept 2000.



# Beta-Carotene may protect against cancer

## ... unless you smoke or drink

adapted from an article by Linda Wang, Assistant News Editor, Katherine Arnold, News Editor

A new study suggests that cigarette smoking and alcohol intake may reverse potential benefits that the vitamin A precursor beta-carotene has on reducing the risk of colorectal polyp recurrence. Colorectal polyps, or adenomas, are benign tumors that may develop into colorectal cancer. The new study appears in the May 21 issue of the *Journal of the National Cancer Institute*.

Two previous randomized trials, which included mainly cigarette smokers, found that beta-carotene supplementation was associated with an increased risk of lung cancer, particularly among those who also drank alcohol. A third randomized trial, which enrolled mostly nonsmokers, found no association between beta-carotene supplementation and risk of lung cancer, suggesting that cigarette smoking and alcohol intake were somehow associated with the adverse effects of beta-carotene.

John A. Baron, M.D., of the Dartmouth Medical School, Lebanon, N.H., and his colleagues looked at how alcohol intake or smoking might modify the effects of beta-carotene on colorectal adenoma recurrence. They examined data on 864 people who had participated in the Antioxidant Polyp Prevention Study. The participants, who were polyp-free after having had previous polyps removed, were ran-

domly assigned to receive a placebo, beta-carotene, vitamin C plus vitamin E, or beta-carotene plus vitamins C and E. Participants completed a questionnaire about their smoking habits and alcohol intake.

Among patients who did not smoke or drink, beta-carotene supplementation was associated with a 44% decrease in the risk of colorectal adenoma recurrence. However, among participants who smoked and also drank more than one alcoholic beverage per day, beta-carotene supplementation more than doubled their risk of adenoma recurrence.

Baron and his colleagues acknowledge that although their study was randomized, alcohol intake and tobacco use were reported by the subjects themselves. "Consequently these exposures bring with them the limitations of most observational analyses, including the potential for measurement error and association with other unknown lifestyle factors," the authors write.

They say that caution must be taken in choosing interventions for large-scale use, particularly when possible interactions with lifestyle factors are not well understood.

Baron JA, Cole BF, Mott L, Haile R, Grau M, Church TR, et al. Neoplastic and antineoplastic effects of carotene on colorectal adenoma recurrence: results of a randomized trial. *J Natl Cancer Inst* 2003;95:717-22. [jncicancerspectrum.oupjournals.org](http://jncicancerspectrum.oupjournals.org)

I understand how you feel about talking about VHL. I find it easier to talk to people in the online support group than I do my family and friends. They are supportive, but those who have or are experiencing anything to do with VHL seem to be more supportive and helpful.

I had a tumor removed from my brain in April and 2 tumors removed from my kidney on the in October. I was really scared about the kidney surgery, but everyone here helped me get through my fears. I knew because of them what to expect, and I found the greatest comfort here.

It is hard to deal with, but if you need someone to talk to, I am here. If I can't help you with certain things, someone here can. I still have my fears, and my down times, but the group pulls me through.

— Loretta G., Tennessee  
[vhlfa@yahoogroups.com](mailto:vhlfa@yahoogroups.com)

## Resources

Joan Borysenko, Ph.D. has a powerfully clear personal vision -- to bring science, medicine, psychology and spirituality together in the service of healing. Trained as both a medical scientist and a psychologist, Dr. Borysenko has gone beyond her traditional academic training and developed depth and breadth in a number of fields including behavioral medicine, stress and well-being, psychoneuroimmunology, women's health, creativity and the great spiritual traditions of the world. Also a licensed psychologist, Dr. Borysenko was co-founder and former Director of the Mind-Body clinical programs at two Harvard Medical School teaching hospitals, now merged as the Beth Israel/Deaconess Medical Center in Boston. These programs were the foundation for her 1987 classic New York Times bestseller *Minding the Body, Mending the Mind*. For books and tapes by Borysenko, see [www.vhl.org/bookstore/wellness.htm](http://www.vhl.org/bookstore/wellness.htm)

Jon Kabat-Zinn, Ph.D., Associate Professor of Medicine at University of Massachusetts Medical Center in Worcester, Massachusetts, teaches

Resources, continued from page 6

mindfulness meditation as a way of managing stress and chronic pain. One way to envision how mindfulness works is to think of the mind as the surface of a lake or ocean. There are always waves, sometimes big, sometimes small. Many people think the goal of meditation is to stop the waves so that the water will be flat, peaceful, and tranquil – but that is not so. The true spirit of mindfulness practice is illustrated by a poster someone once created of a 70-ish Indian yogi, in full white beard and flowing robes, atop a surfboard and riding the waves off a Hawaiian beach. The caption read: “You can’t stop the waves, but you can learn to surf.” For books and tapes by Kabat-Zinn, see [www.vhl.org/bookstore/wellness.htm](http://www.vhl.org/bookstore/wellness.htm)

Deb Hogan (right), a singer in the Boston area, was diagnosed with VHL in 1998. Singing songs of faith gave her renewed strength. She decided to make a CD to share these songs with others going through crisis. “I have had to face my own mortality in the last few years, and that’s been very hard. But



my faith has given me peace and given me joy.” Forever In His Care is Deb’s first CD, a collection of songs to help others overcome fear and gain contentment. They are songs of her Christian faith and hope, especially focused on a path away from worry and into trust in God. CDs are available from [www.walkoutofworry.com](http://www.walkoutofworry.com)

## Genetics Home Reference Makes Genes, Chromosomes, DNA Easily Understood

(Bethesda, Md.) – When you hear “gene map,” do you think it’s a guide to finding the nearest Gap store? Are you the kind of person who thinks that “genetic markers” are sold at office supply stores?

Now, thanks to the National Library of Medicine (NLM) you can find answers to your genetic questions. With the click of a mouse you can go the NLM’s newest consumer web site, “Genetics Home Reference,” at <http://ghr.nlm.nih.gov>. Genetics Home Reference joins Medlineplus.gov (the consumer site for general medical information) and Clinicaltrials.gov (the site that lists clinical research trials) in the lexicon of NLM’s consumer medical web sites.

“The American public is increasingly turning to the Web for medical information,” said Donald A.B. Lindberg, M.D., director of the National Library of Medicine. “The launch of Genetics Home Reference was a logical step in making genetics and its relationship to disease more understandable to the general public,” said Lindberg.

“Knowledge about genetics is vital for a true understanding of many diseases,” says Alexa T. McCray, Ph.D., director of NLM’s Lister Hill National Center for Biomedical Communications, the organization responsible for creating this innovative website. “Often, individuals need to make life-altering decisions because of their genetic background. We hope that Genetics Home Reference can help guide them as they make their medical choices.”

The target audience is the general public, and the language is written at the high school level – for those who remember “a little from their high school biology course.” A quick refresher course is available by clicking the “help me understand genetics page” which talks about, for example, how genes can be turned on and off in cells, what it means if a disorder seems to run in a family, and the principles of gene mutation. Explanations are written in simple and understandable English.

If you have questions about a specific disease you browse either by disease/condition or by gene. If you type in Alzheimer’s disease, for instance, a page appears where the information is written in a question and answer format. You’ll find out how people inherit Alzheimer’s, the symptoms, and what treatments are available. There’s also a geographic listing of genetic counselors and information for care-givers. In addition, you can easily find details on the specific genes related to Alzheimer’s.

Other features are a glossary of genetic terms, links that take you to clinical trials related to the disorder you’re searching, and more advanced genetic information. Genetics Home Reference will be adding genetic diseases on a regular basis and the information will be updated as needed.

The National Library of Medicine is a part of the National Institutes of Health, an agency of the U.S. Department of Health and Human Services.

# Support and Awareness

By Karen A., New Jersey

On March 27th, 2003, I held a very easy fundraiser for VHL Family Alliance. I decided to hold a Pampered Chef Party! This was only the second one I had ever thrown and it was a lot of fun! It's a home sales party, like Tupperware or Mary Kay -- and let me just tell you that it's almost always a fun group of women looking for a couple of hours away from responsibility.

A Pampered Chef Hostess came to my home and set up her table and items for sale. Then she showed us how to create simple dishes using everyday items from the fridge or grocery store. After her demonstration she served the food to us, played some fun games with prizes, and asked us to look through the catalog.

While this was a fun night for all, full of eating, drinking and shopping, I turned it into a wonderful night with a very important purpose. The proceeds from my show went directly to the VHL Family Alliance.

In a two hour show, coupled with donations, I was able to raise over \$800 for VHL. Not all of my friends and family could attend so I passed out catalogs to my friends who live locally and sent an e-mail to those who live out of town. I reached over one hundred people via e-mail and close to one hundred people locally. Some people didn't want to purchase anything, so they sent me a donation. Others could go to the Pampered Chef website and see what there was to purchase, then e-mail their order to me. They sent checks or called with credit card numbers.

It was a phenomenal success and I encourage everyone reading this article to contact someone locally who does a home-based show. There are tool shows for women, pajama shows, children's toy shows, Tupperware shows and even lingerie shows! The possibilities are endless and all of these organizations have fundraising departments.

My son is going to be 7 in May. He was diagnosed at 5 years old with an endolymphatic sac tumor (ELST)<sup>1</sup> on his right side. There are two things I can do for him. I can be certain he gets the best medical care there is for him, and I can raise money for VHL to contribute to research. The endolymphatic sac tumor is the most rare and least researched tumor of all VHL tumors. My son is one of the youngest ever to be diagnosed.

I will not sit idle and be a victim, I'm taking action. I'm working for a cure for VHL and a better life for my son. You can too. Please, I encourage you

Alex



to pick up the phone and call anyone you know who has a home show business. You will not only raise much needed money, but you will raise awareness, and for that there is no substitute!

1. See Screening for ELST, *VHLFF*, September 2002.

## Ask the Experts

Question: Do you have any statistics on how many young people (up to 18) may experience significant problems with vision? -- Gillian C., Australia

Answer: As for statistics on young people, there is difficulty in collecting such data because we do not have the denominator for these numbers. We have some counts, but we have no idea what percentage that might be because we don't know how many people with VHL there are in total.

From our own studies here among more than 400 patients with VHL, the number of young people who are severely affected is low. In fact, the average visual acuity of all our cases is 20/25 in one eye. About 11% of our patients have optic nerve lesions, 1% have them in both eyes. Approximately 8% have enucleation or loss of one eye.

We see all patients as part of the screening protocol, not just when they have severe eye problems, so we probably have a better perspective on the general situation than most eye doctors. While children may develop eye problems at early ages (2 or 3), if they are treated appropriately they don't usually get into trouble until sometime in their 20's. We are always hoping to reduce the severity of eye disease in VHL. We have started a trial with an anti-angiogenic agent.

-- Emily Chew, M.D., National Eye Institute, National Institutes of Health, Bethesda, Maryland



# Uninsured are at a Disadvantage

— from JNCI Cancer Spectrum, Oxford University Press, <http://jncicancerspectrum.oupjournals.org/>

Almost 41 million people in the United States lack health insurance, more than the populations of Texas, Florida and Massachusetts combined. Uninsured individuals are more likely to have poorer health and die prematurely than those with insurance. Their families and communities are also affected. Communities with large uninsured populations are more likely to reduce hospital services, divert public resources away from disease prevention and surveillance and reallocate tax dollars to pay for uncompensated medical care.

So says the new report *A Shared Destiny: Community Effects of Uninsurance* from the National Academies' Institute of Medicine. This report is part of a series examining the consequences of being uninsured for individuals, families, communities and society as a whole.

The report concludes that Americans without insurance have poorer health and are more likely to die prematurely than people with insurance. Providing health insurance to the uninsured could improve their health and extend their life expectancy, the report concludes. Moreover, increased health coverage could reduce health disparities among racial and ethnic groups.

Among people with breast or colorectal cancer, there was a 50% increased risk of death if the patient did not have insurance. This increase in risk was largely attributed to delayed diagnosis because the uninsured are less likely to receive timely screening tests such as mammograms, Pap tests, and colorectal screening. Uninsured cancer patients often have cancers that go undetected until a fatal stage.

Lack of insurance also affected the type of treatment a cancer patient might receive. For example, uninsured women with breast cancer were less likely to receive breast-conserving surgery than women with private insurance.

The report also looked at the impact of insurance on diabetes, HIV and AIDS, health and kidney diseases, mental illness, traumatic injuries, and heart attacks. Conclusions were drawn from health outcomes of roughly 30 million Americans who were uninsured and between the ages of 18 and 64.

The full report is available for free download from the National Academies at [www.nationalacademies.org](http://www.nationalacademies.org)

The Robert Wood Johnson Foundation is heading some initiatives to help at 1-202-572-2928 or [covertheuninsuredweek.org](http://covertheuninsuredweek.org)

Learn more about your rights under the Health Insurance Portability and Accountability Act (HIPAA), designed to protect workers who are changing or losing jobs. See [www.ahrq.gov/consumer/insuranc.htm](http://www.ahrq.gov/consumer/insuranc.htm)

The U.S. government Agency for Health Care Research and Quality (AHRQ) provides consumer information on health insurance options and choosing a quality health plan. See [www.ahrq.gov/consumer/hlthpln1.htm](http://www.ahrq.gov/consumer/hlthpln1.htm)

The National Organization for Rare Disorders (NORD) at 1-800-999-6673 or [www.rarediseases.org](http://www.rarediseases.org) administers access to some prescription drugs for people in need. The website at [www.benefitscheckup.org](http://www.benefitscheckup.org) helps people over 55 find assistance in paying for prescription drugs.

Get information on your state's Children's Health Insurance Program (CHIP) and contacts for Medicaid administrators in your area at 1-877-543-7669 or [www.insurekidsnow.gov/states.htm](http://www.insurekidsnow.gov/states.htm)

The U.S. Department of Health and Human Services' Bureau of Primary Health Care has information on public programs for the uninsured in your area. See the web at [bphc.hrsa.gov](http://bphc.hrsa.gov) or 1-301-594-4110

## Nashville Regional Meeting

June 21, 2003

St. Paul's Episcopal Church,  
Franklin, Tennessee

Registration: \$25 per person

Y' All Come, now, y'hear?

## Local Activities

Several of the chapters held special activities, especially during VHL Awareness Month in May.

Terry W. met with the University of South Carolina genetic counselors' program in March. It's a post graduate program. "I get an hour of their class time to talk about VHL and show the VHL video. The students are seniors in the program, and are from all over the US. Recently one young lady was from as far away as Washington State. She had graduated from Washington State University, and another was from Michigan, where she had attended the University of Michigan. I've done this for 6 or 7 years. In the beginning I didn't have the video. It has been a nice help." The VHL video is available from the office, 617-277-5667.

The California state meeting was held in Sacramento in April. Dawn Cerf reports, "Dr. Ralph de Vere White, the Director of the U.C. Davis Cancer Center, discussed his work on VHL, and we were able to ask lots of questions. As a side note, one of his specialties is prostate cancer. His research has revealed that the much higher incidence of prostate cancer among the African American population is correlated with their high dietary intake of BBQ meat. That's something to think about. We noticed that no one ate BBQ at lunch.

"After our speaker left, the group talked for hours. We had so much to say to each other. One of the attendees had been officially diagnosed with VHL only days before the meeting. Needless to say, he was delighted to meet everyone and receive lots of good information. We didn't want to overwhelm him, but he wanted to soak it all in. Best of all, our conversations were sprinkled with lots of humor and laughter. We were able to discuss a serious topic and enjoy ourselves at the same time. There was a deeper level of bonding going on that happened because our meeting was smaller than usual."

A few members met in Phoenix, Arizona, with Peggy and Don Marshall during their vacation there



Three generations of women with VHL, Rhode Island

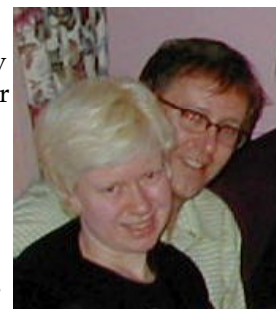


California meeting, University of California Davis, in Sacramento, March 2003. Collage by James Thompson.

in May, celebrating their 42nd wedding anniversary. Happy Anniversary to Don and Peggy!

A few members met in Augusta, Maine, with Joyce Graff, following the American Cancer Society Living with Cancer Conference in May at the Augusta Civic Center.

A group of Rhode Islanders met for a delightful dinner with Joyce on May 21 at a restaurant in Providence. In particular we had two very animated women with similar brain tumor histories, very engaged in sharing stories and local resources and strategies for caring for their health. We also had one family with three generations of women with VHL — ages 31, 51, and 72 — all eating and joking with us at the table, which was very inspiring to the young newly-married couple.



Amy and Steve, Rhode Island

The Massachusetts chapter held a fundraiser in conjunction with Fajita's & 'Rita's Restaurants in May 2003. We invited our supporters to have dinner, and Fajita's & 'Rita's donated 20% of the dinner check to VHLFA!

The New York chapter is meeting June 1 and will report their activities in the next newsletter.

Chile now has eight family groups with VHL, including a new one identified in April. The medical community is becoming more aware of VHL, and the Catholic University in Santiago is developing a coordinated center for clinical care. Chairman Pierre Jacomet is being interviewed June 3 on television with a live audience of 500 people. He is a well-known writer and philosopher in Chile, and he will use ten minutes to talk about VHL.

Happy 42nd Wedding Anniversary to Don and Peggy Marshall!

# Ask the Family: Life with VHL

Question: I'm writing a report and I need to know what a day is like when you have VHL. -- Carla V.

Answer from Amélie, France: Ups and downs, as in life in general. A malady is like a person -- you have to get to know it in order to live with it. I am grateful for all the good information in the Handbook.

Answer from Cathy C., New York: My life with VHL....I work 45 to 50 hours a week running a store. I'm on my feet 10 hours a day in constant motion at work and then I come home and mow the lawn. If I'm working late I walk five miles with my walking buddy in the morning. I feel I'm very lucky to have the life I have, and I enjoy every minute of it. I'm healthier than I've ever been in my life because before VHL when I didn't feel good, I just lived with it. Now that I know I have VHL I go to the doctors and do something about it. I'm living a much healthier lifestyle and I take care of myself. There are good sides to VHL, we just need to look for them.

Answer from Babs M., Australia: Well for me, there are good and bad days. I can't scoot on my scooter at the moment, because of balance but I'm not bumping into walls anymore, which is a great relief to my family. I am slowly regaining function after my brain surgery. Nor am I spending so much time picking myself of the floor. Looking at the ceiling from the floor really helps you get a good perspective of what things look like to a baby or small child.

I'm having a lovely Mother's Day here today, despite the fact that I woke up with a really stiff and sore left arm and shoulder, and couldn't meet people for breakfast..(Left over from a bad inline skating injury I sustained a few years ago.) The shoulder injury has nothing to do with VHL, but it's all connected. I thank God for the invention of the rice sock, which is useful for managing pain without drugs. It's a sock (actually it's a tube sock from an airplane trip) that I fill up with uncooked rice. I heated it up initially in the microwave for about 1 minute on high. But now I have used it so much, that I only need to heat it up for about 45 seconds. Then just place it on the part that is painful. If it's too hot I put another sock on top. I've found it a bit safer than a hot water bottle, which can be a bit dicey on the face or ears.

I meditate as well as do stretches every morning when possible, which helps with the mind-set for the day.

I try to hang out with positive thinking people as much as possible. Which means that I don't always like to hang out with myself! I also have a pretty warped sense of humour I have been told, that has helped me get through a lot of things.

I keep myself as busy as I possibly can without getting too tired. I don't have as much stamina as I used to have before my brain surgery, but I still try to keep up my pretty hyperactive pace.

I guess you could say that I take the lemons I have been dealt in life, and make lemonade, lemon spread, lemon meringue pie. I eat lots of veggies and fruit. Make sure I keep up with my doctor and dentist appointments, plus my acupuncture treatments.

In some ways I am even more active than I was before. I have found there are pluses and minuses with VHL. And I have been and am very blessed to have some very inspiring people in my life.

Answer from Jennifer K., Australia: My life with VHL ... I have had several spinal surgeries that have left me with some deficits, but I'm generally doing well. At the moment my day consists of gentle Yoga (for flexibility and strength), volunteering at the Art Gallery and the Aquarium (to keep my brain active), going to the osteopath (nerve and tissue re-alignment), having a massage (for muscle and pain relief) and acupuncture (haven't tried this before, but I am doing it hoping it will help my neck movement and proprioception. That's your ability to know where your legs and arms are. Sometimes I have to look over at them or move them to be sure where they are! Fortunately, they are usually right where you left them!

We have just moved to a new town and I'm still doing my medical shopping but I keep up with my medical surveillance and I'm pretty much on top of it now. This town doesn't have all the medical experience and knowledge needed. That's why the Alliance's web site is so helpful. I give that address to doctors and anyone else dealing with VHL, so they can get accurate information. It's great!

I feel it is important to equip oneself with the skills needed to handle or control to the best of one's ability one's own situation. And everyone's needs are different. That's another reason the Family Alliance is so helpful. We help each other and one can talk with someone else who at least knows what VHL is and can help because they've been there.

# Tissue Donor Registration Form

I, \_\_\_\_\_ wish to register myself (or a dependent minor) as a VHL tissue donor with the Brain and Tissue Bank for Developmental Disorders at the University of Maryland, Baltimore. This donation grants permission for the Brain and Tissue Bank to make every attempt within its means to coordinate recovery of surgically removed tissue of the above named donor. Further, if death should occur, I (☐ do or ☐ do not) hereby grant permission for recovery of brain and other tissues. All tissue is donated for the expressed purpose of furthering the research of von Hippel-Lindau disease.

Donor	Next of Kin
Name _____	Name _____
Address _____	Address _____
City _____	City _____
State/Province _____	State/Province _____
Zip/Postcode _____	Zip/Postcode _____
Phone evening _____	Phone evening _____
Phone daytime _____	Phone daytime _____

Donor's Date of Birth \_\_\_\_\_ Sex \_\_\_\_\_ Ethnic Group \_\_\_\_\_

Brief Medical/Family History (add paper as necessary) \_\_\_\_\_

If you (the Donor) are not diagnosed with VHL, are you the parent/relative of someone who is?

☐ Yes ☐ No. Describe the relationship \_\_\_\_\_

Signature of Donor or Legal Guardian: \_\_\_\_\_ Date \_\_\_\_\_

Feel free to include any further relevant information.

Please mail to:

Brain and Tissue Bank for Developmental Disorders  
University of Maryland at Baltimore, Department of Pediatrics  
655 West Baltimore Street, Room 10-035 BRB  
Baltimore, Maryland 21201

Questions?

Call Christine Wade  
Toll free U.S./Canada: 1-800-847-1539  
Tel: +1(410)706-1755; Fax: +1(410)706-0020

## Important -

In case of surgical emergency, or in case of death, please notify the Tissue Bank immediately, any time of day or night, on 1-800-847-1539. Tissue not recovered within 24 hours cannot be used for research.

Thank you!

## Enucleated Eye Tissue Needed

The U.S. National Eye Institute is doing a study using angiogenesis inhibitors to control eye tumors without laser or cryo. You can help with this study by donating enucleated eye tissue for research. If you are having your eye enucleated, please have your surgeon contact Dr. Chew prior to the procedure to arrange tissue transfer:

Dr. Emily Y. Chew, Natl Eye Inst  
NIH, Bldg 31, Room 6A52  
31 Center Drive MSC 2510  
Bethesda MD 20892-2510  
W: 301-496-6583  
Fax: 301-496-2297



# VHL Tissue Bank

## An Essential Aid for VHL Research

We are constantly striving to increase the level of VHL research. Once considered only "an obscure medical curiosity", VHL is becoming one of the most important diseases in the study of cancer. The discovery that the VHL gene is mutated within the cells of clear cell renal cell carcinoma in the general population should tend to raise VHL research to a new level. While it is estimated that there are only about 7,000 people with VHL in the United States, it is estimated that more than 25,000 people will develop clear cell renal cell carcinoma in the United States each year.

As the level of VHL research increases, the need for VHL tissue for research also increases. It is here that we can help. The VHL Family Alliance, in conjunction with the Brain and Tissue Bank for Developmental Disorders of the University of Maryland, is proud to announce the establishment of a VHL Tissue Bank. Tissue donated by VHL patients will be held in the Tissue Bank until an approved research project has need for it.

If you have been diagnosed with VHL, and are

contemplating surgery, you can help the research community by donating any surgically removed tissue to the VHL Tissue Bank. All cost and arrangements for recovery and transfer of tissue will be taken care of by the Tissue Bank. If you would like to help the VHL research effort, please fill out the Donor Registration Form and mail it today. All information will be treated in the strictest confidence. Pre-registration makes the process simple in the event of surgery. Simply contact the tissue bank, give them the name and contact information for the surgeon and the date of surgery, and the Tissue Bank will make all the necessary arrangements. Even if you are not already pre-registered, arrangements can be made by contacting the Tissue Bank.

Give a gift that only you can give, and help promote research on VHL. Researchers interested in access to tissue on file should submit research proposals to Joseph Verdi, Ph.D., VHL Family Alliance Research Committee, 171 Clinton Road, Brookline, MA 02445, or e-mail to [info@vhl.org](mailto:info@vhl.org)

To register, please fill out and submit Registration Form on page 12, or online at [www.btbankfamily.org](http://www.btbankfamily.org)

# Welcome to the Tissue Bank

It is an honor to us to be the official Tissue Bank for the VHL Family Alliance. We can assure you that without exception all tissue received through referral from the VHL Family Alliance will go exclusively for the study of VHL unless other arrangements have been made through communication with you.

We are always encouraged by working with groups such as yours which evidence such a deep commitment to further research on the cause and treatment of the various developmental disorders.

I believe that The Brain and Tissue Banks for Developmental Disorders will be of great service to those members of the VHL Family Alliance who have concluded that the greatest gift that they can make to further medical research is the donation of tissue either at the time of surgery or at the time of their death. The availability of such tissue will encourage new research studies that are currently not feasible and will bring us closer to the day that disorders such

as VHL are no longer a mystery.

A tissue donation program works through the combined efforts of support groups, their membership and the research community. We have found that this sort of cooperation fosters a sense of alliance, comfort and hope in all the diverse individuals who are working towards the same goals. We hope that you too find this to be true.

Please feel free to call either Dr. Zielke or myself at any time if questions arise. Our phone, 1-800-847-1539, is answered on a 24-hour basis. Thank you once again for your confidence.

Tissue Bank at the University of Maryland  
Phone: 1-800-847-1539 or +1 (410) 706-1755  
Fax: +1 (410) 706-0038  
E-mail: [btbumab@umaryland.edu](mailto:btbumab@umaryland.edu)  
H. Ronald Zielke, Ph.D., Director  
Robert Vigorito, Tissue Coordinator  
Christine Wade, Project Coordinator

The Brain and Tissue Bank accepts tissue from a number of countries including U.S., Canada, Mexico, and Australia. If there is not a separate research project in your country, please inquire about transfer to the BTBank. Additional forms may be required when crossing national boundaries.

In Britain and Ireland, call Prof. Eamonn R. Maher  
Med. Genetics, University of Birmingham  
+44 121 627 2741; Fax: +44 121 627-2618  
E-mail: [E.R.Maher@bham.ac.uk](mailto:E.R.Maher@bham.ac.uk)

In France, call Dr. Stéphane Richard  
Oncogenetics, Kremlin-Bicêtre Hospital  
Phone/Fax: +33 (1) 49 59 67 28  
E-mail: [stephane.richard@kb.u-psud.fr](mailto:stephane.richard@kb.u-psud.fr)

In Germany, call Prof. Dr. Hartmut Neumann  
Medizinische Universitätsklinik, Freiburg i Br.  
Tel. +49 761 270-3578, FAX -3778  
E-mail: [neumann@mm41.ukl.uni-freiburg.de](mailto:neumann@mm41.ukl.uni-freiburg.de)

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Editor: Joyce Wilcox Graff, 1-617-232-5946 (eve)  
Adviser: Debra L. Collins, M.S., U. Kansas Med. Center, 1-913-588-6043  
Internet website <http://www.vhl.org>  
171 Clinton Road, Brookline, Massachusetts 02445-5815 U.S.A.

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All members receive the VHL Family Forum quarterly. Check here ☐ if audio version is needed☐ New member ☐ Renewal (Dues \$25 per household, \$35 per professional) \$ \_\_\_\_\_☐ I cannot afford dues at this time, but please accept my donation (any amount) \$ \_\_\_\_\_☐ Please send me \_\_\_\_\_ brochures about VHL Research Fund to share with others

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\_\_\_ VHL Cookbook, fun and delicious recipes from members worldwide @12.00 ea \$ \_\_\_\_\_

\_\_\_ VHL Lap blanket, all-cotton woven thermal throw @ \$40 each (brown &amp; tan) \$ \_\_\_\_\_

\_\_\_ Caring T-shirts (brown print on tan shirt) Size XL @\$10 ea \$ \_\_\_\_\_

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Canadians, please send donations to: Canadian VHL Family Alliance,

4227 Hamilton Road, Dorchester, Ontario, N0L 1G3 Canada

## 6<sup>th</sup> Medical Symposium on VHL in Japan

The first major medical meeting on VHL to be held in Japan will be at the Kochi City Culture Plaza in Kochi, May 20-23, 2004. We have designed the meeting over a four-day period to meet the needs of attendees with different medical backgrounds.

Thursday, May 20: Basic Science Day, and Opening Ceremony

Friday, May 21: Clinical Session, Genotype/Phenotype Alignment, and Dinner

Saturday, May 22: Clinical and Basic implications

Sunday, May 23: Summary, Open Questions, exchange of ideas between patients and doctors

This meeting is to foster interaction among researchers and clinicians. Families are welcome to attend, but should know that no attempt will be made to constrain the language, so it will be pretty technical. Thursday in particular will be a sharing of the state of research on the workings inside the cell, so the language may be difficult even for clinicians. Sunday's meeting is the portion designed specifically to include families.

Kochi is a long, fan-shaped area stretching from east to west, which takes up almost all the southern half of Shikoku Island. It has a warm, subtropical climate. Kochi City Culture Plaza is a community education and cultural center, with meeting rooms for conferences like ours.

Kochi Castle was built in 1600 by Kazutoyo Yamanouchi. The castle tower and gates are rich in the culture of the area. From the top of the castle you can see out over Kochi city.



Katsura-Hama is a lovely beach, well known as the most beautiful sight-seeing point in Kochi. You can enjoy a very blue sea and a picturesque moon. The Yosakoi dance festival features more than 15,000 colorful dancers performing in the streets to the accompaniment of naruko, a kind of instrument unique to Japan.

Hotel rooms have been blocked at Hotel Nikko Kochi, Kochi Daiicchi hotel, and others. More details will be made available in future newsletters, and on the web at [www.vhl.org/conf2004](http://www.vhl.org/conf2004). Those interested in presenting at this conference should contact the Department of Urology, Kochi Medical School, at [vhlsympo@kochi-ms.ac.jp](mailto:vhlsympo@kochi-ms.ac.jp)  
See you there!



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