

Coping with VHL

Gary L. Wood, Psy.D., Florida, from his talk at the Florida Regional Meeting in Tampa, January 2004

It's always pleasing to be around people like those in this room, with a passion for their work, and a vision ... all of us, working together to address the challenges that affect our lives.

I am a licensed psychologist in the state of Florida. I own and operate a consulting firm here, and interestingly I do very little work dealing with patients who have physical health problems other than my own. I spend most of my time administering employee assistance programs for about 30 companies. They are based here, but they are all around the country. My primary work is in evaluating employees and family members who are faced with behavioral health conditions – primarily emotional problems or lifestyle problems. We do in our organization provide educational activities that focus on building resilience and reducing stress and so forth.

I am here to talk about the psychological aspects of this illness, and quite frankly I don't know that we can say a whole lot about the science of the psychological aspects of VHL because we don't have the data to offer. There was a talk by Dr. Joseph Locala at the VHL meeting at the Cleveland Clinic. But in the medical literature I could only find one article on the psychological issues of VHL, written in 1964, the year I was diagnosed with VHL, 40 years ago.

I have a lovely daughter who has this disorder also. And like you and your family members, we are trying to stay on top of VHL -- monitoring and doing good medical surveillance.

But first, please tell me what you would like to talk about on this topic.

1. At what age would you talk with a child who has been diagnosed with VHL.
2. How do you deal with the anxiety of waiting for test results?
3. What are the medical indicators of the progression of this disease in specific body parts. How do you deal with that?
4. How would you address an individual who won't go for testing? My grandfather was the one who

had VHL, and my father was from a very large family, but very few of his relatives have gone for testing.

You have to work out a way of going from putting your head in the sand to wearing it as a badge of courage. Good questions.

I think that the literature that you have to draw on to address some of your questions is the literature that we know best, and that is dealing with loss. We don't necessarily know a lot about the particular behavioral manifestations that occur as a result of a diagnosis of VHL. But we do know something about how people react from a psychological standpoint in terms of facing loss. There have been some fairly good studies in cancer patients and how they react.

Now, I think it is important that we remember that VHL is primarily a physical disorder; it is not a psychological or a psychiatric disorder. And as people begin to try to gain awareness and insight into the nature of their condition, you have to be careful about the differential diagnosis in obtaining accurate and reliable information, or you can drift into speculation -- where every person has their own theory about what might work, but little real data. We could probably come up with some fascinating psycho-babble to try to help people a little bit. But I think we have to pay close attention to each case and obtain accurate and reliable information to help prepare the individual for what kind of loss, what level of loss, he or she will experience.

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We do know from looking at the literature that some people fare better under loss than others. And we do not always know what are the prerequisites for doing better with loss. We know that certain individuals do better in adversity than others. And how do you identify those who do better than others? It's just hard to say. I think that this is a line of research that would be useful in studying VHL patients.

But I think that when you are trying to face some of your questions — How to talk with children, how to face family members who may be in denial — it's a good idea to remember for yourself and for them that they are facing a loss.

Now when you think about a loss, what comes to your mind? Death. Yes, that is the most stressful event that any of us can experience. What else? Loss of relationship, lack of interest, religion, faith, work, loss of independence. Yes. This study that was published in 1964 by a group of psychologists and psychiatrists is a study of a family of twenty-four people in San Francisco, California. The study focused on some of the behavioral characteristics of the family members. And some of the things that were noted are things that we see in loss — denial, anxiety, and avoidance.

Whenever someone experiences a loss, the first experience they have psychologically is shock. If you had a physical injury, your body would go into shock. When you learn that you have this diagnosis and you begin to get more information, you begin to experience a loss. And that is a normal reaction, it is not a pathological reaction. Loss is a normal psychological experience in human nature. And to experience shock and disbelief is a normal reaction. As a matter of fact, if you don't experience shock, then we have a problem. Because then we would see you develop a lack of compliance with further medical evaluation and treatment, affecting the neurotransmitters chemicals. Your brain is a three-pound bag of juice. What you think and what you do have an impact on your chemistry. So people can negatively affect themselves if they don't handle the stages of loss properly.

As parents, as employers, as caregivers, when someone experiences a loss, what do we do? Console. Get angry. Withdraw. Feel sorry for ourselves, be angry, fearful, have no motivation at all. And these are common reactions — not only on the part of patients, but these are reactions that occur on the part of caregivers, particularly if this is a loved one. It is important to understand that the initial stage of shock will subside with time. But you can accelerate the adjustment by offering support.

What is support? Listening. How easy is it to listen when you think that your daughter or your son or your wife has a life-threatening illness?

Hard. Very hard. Unless you have some understanding yourself. I'm sure you are naturally compassionate, and you're a caregiver type yourself, but even the best of us, if we are going to try listen, we should first try to educate ourselves. This is where the value of organizations like this Alliance comes in.

In psychology and in medicine we try to get people to become their own doctor, so to speak. You want get to the point where you are inquisitive and informed. If I am informed, I can listen better. I would be quite able to listen to any one of you about your VHL condition if you came to me as a patient. I think any of you would believe I would understand you, right? It's because I have information about VHL.

And we as parents, we as caregivers, are much more able to help answer the questions of a child, to know what to do with a child, or to hear those family members who feel ashamed of their illness, or guilty that they passed it on to their grandchildren — all the shame and guilt that often comes with a genetic condition. You are more likely to intervene constructively with these people if you have an objective understanding of what's happening.

There's a difference among information, knowledge, and wisdom. Those are different things. And everyone has to address their questions at the level they are most comfortable addressing them, and with whom they are most comfortable. But I can tell you that loss, while it is a normal reaction, can also become malignant.

We are very fortunate to have people who have done research on loss. Dr. Elizabeth Kübler-Ross, who wrote the book *On Death and Dying* and has had a great influence on the grief literature and research on grief, has done a wonderful job of helping us understand the various stages of loss — beyond shock and disbelief and sadness and anger and guilt and — relief. It takes a long period of time to deal with these things.

I was diagnosed in 1964. I discovered that I was blind in my right eye at age 10. Coming from a blue-collar community where there was not much medical care available, I did not know why I had lost the sight of my right eye when I was 10 years of age. When I was 17-18 years old and a very good athlete with just one eye, I discovered for the first time that I would not be able to drive an automobile or shoot a basketball again or chase good-looking girls. It was pretty hard on me. But I was competitive. And I received good support from people I didn't even know cared about me — school teachers, coaches, ministers, a loving family, and a counselor who understood something about grief. Somebody to just sit and listen. These things were very helpful to me at that time.



Gary
Wood
speaking in
Tampa

Out of my own faith – I'm one of those individuals who have a Protestant ethic that is non-dogmatic – I really believed that something positive could happen as a result of this experience. I was fortunate to learn about books you could read by way of audio tape. At that time they were disks – Talking Books, we called them. And the first book I read was the *Introduction to Psychology* – a textbook – they just sent it in the mail. And I said, “I think I want to be a psychologist.” I liked helping people, I was a team leader, and all that sort of thing.

And as I look back on my life and I have studied personalities and over the long haul, I do think that while having personal strength is important in being able to pursue goals, it's critical when you are faced with a limitation – a loss. Having support around you, people who love you, people who can provide you with accurate and reliable information – like this morning, Joyce was providing us an update on some of the research. I'm going to leave here better equipped, aren't you guys? And listening to the experiences of the people here who are participating in some of these Phase 2 clinical trials – this is very helpful, supportive information.

So I think before you start talking with someone, it's good to **get accurate and reliable information**. If your child has been diagnosed with this disorder, you have to deal with your own feelings of loss first. And that means you having someone to turn to, who can support you with accurate and reliable information. Someone in this room, someone on the hotline or the online discussion.

But know that even with the best information, we are still going to feel a loss. We live with a life-threatening illness. Using the science of risk management in medicine, we can help ourselves. The Europeans have brought to us the knowledge of **harm reduction technology**. It helps us understand that there is no one absolute way to do things. You identify a disorder, and you identify the risk factors that accelerate that condition, and you intervene on those risk factors, whether they be physical risk

factors, behavioral risk factors, or whatever. The objective is to reduce harm.

I think that **grief counseling** is a good idea for any of us who are looking for ways to communicate with our family members. It's probably the most important if all the things that I looked at in preparation for this meeting. I hope you will take away from this talk an understanding that loss is a normal process. If you are trying to help yourself or a family member, then grief counseling is a good idea.

I have had to face this myself. I face it every day – we all do – especially when you get an ache or a pain or a sensation – hmmm, is this a tumor? I have a pain in my back – is this a tumor? Feeling a little unusual – do I have an adrenal gland tumor?

What's going on here? If you're neurotic like I am, you start looking into it. I'm an information nut. I get a test as often as I can, and I make sure that my daughter gets them, and she's probably neurotic by now too. Not really, she's pretty easy about them.

At this point, his daughter teasingly interjected that Gary does still play basketball, and that good-looking girls now chase him!

I think that I would encourage all of you to learn as much as you can about grief and loss. Dr. Kübler-Ross has written a lot about it. I think it would help you in addressing ways that you would want to communicate with family members as well as deal with yourself. I also think that it will help you to look at your own grief process – your denial, your anger, your aggression, your desire to try to make things happen when you can't make them happen, and the resulting feeling of stress and frustration. We do know that extraordinary anger can compromise one's immune system.

We also know that positive psychology such as optimism, is good for your health. We know that looking at things realistically, finding meaning in adversity, and trying to make do with what you have, can cause secretion of hormones such that when you look at the brain with a functional MRI, you see the parts of the brain that are associated with resilience and coping light up! It's beautiful!

So as you consider looking at your own grief, and those children you may have, or those children that you do have, keeping focused on ways that you can get your frustrations out, talking with someone. For me, usually it's a friend. Although I have been through therapy myself and I found it very useful.

Now, grief is a process that takes time. Some of the information that I have collected suggests that it takes a period of several years after each loss. And in our situation there are many losses, aren't there? And they can come regularly. There are periods of time between our losses. I lost my sight. That was a very specific loss related to VHL. Some of you have not lost anything physically at this point, but you

may be concerned about losing something, and often the anticipation of loss can be as real as the loss itself.

There are specific psychiatric conditions that can develop independent of VHL and I'd like to touch on some of these. VHL does not cause these things, but if you have them you do want to know about them.

Major depression is a clinical disorder that is characterized by recurring feelings of sadness and loss of pleasure and enthusiasm. Individuals who have this disorder have had it for more than a month, and they also have trouble concentrating, they have trouble continuing to participate in activities that they normally participate in. Essentially they have a decrease in their energy. They often have sleep difficulties, particularly early morning awakenings. One's thinking is negatively impacted – you are not able to get projects done as you used to get them done, your interpersonal relationships are challenged, you may lose intimate desire, you may become more irritable or agitated. In some instances people who suffer from this depression may act as though they don't have it. We call these smiling depressions.

The bad thing about major depression is that it feels really bad. The good news is that it's very treatable. We have very effective treatments for major depression and we can clear it up quickly with proper diagnosis and proper treatment.

Another disorder that is quite common particularly in people with medical conditions like ours are **anxiety disorders**. Anxiety disorders can be characterized by what we call generalized anxiety which is a free-floating sense of anxiety throughout the day. You may have periodic panic feelings, but most of the time there is a generalized anxiety, a grand sense of doom, you're a worry-wart and everybody knows it and you know it. And you can't determine if you're anxious because you were abused as a child, or your mother didn't treat you right, or you're dying, or what. You just feel overwhelmed, as if you can't cope. But you keep going.

Usually people who have anxiety are what-if thinkers. They never answer the question, they just keep thinking what-if-this and what-if-that. Anxiety can also result in phobias, when you avoid people or places. You can develop anxious associations with doctors or hospitals or other places. You feel they are unpleasant, and want to get away from them. We have successful treatments for anxiety.

Panic attacks are another disorder that can occur. Panic attacks are physical sensations that you have that may last 10-30 minutes and then they subside. But you feel like you're going to have a heart attack when you have one of these. You may even go to the Emergency Room and they don't find

anything and they tell you it's all in your head. These kinds of conditions are very real and very treatable with the right diagnosis and appropriate treatment.

Some individuals when they lose something have a **bereavement reaction**. This goes beyond normal grief. The person develops symptoms of major depression. They lose interest in life. It's all associated with a loss. You have not worked through the loss, and now you have developed an emotional health disorder as a result of the loss.

There are many other conditions that you can develop, but these are some of the more prominent ones. If you have persistent feelings of sadness or anxiety, or if you had a loss and you are unable to work through it, it is good to consult with a psychologist or a psychiatrist or a qualified mental health professional in your area so that you can determine if you have one of these disorders, so that it does not negatively impact your further follow-up with VHL.

With VHL losses can become so compounded that you don't have time to breathe and mourn and instead you go into denial. A little denial is a good thing, and is sometimes a characteristic of resilience. But if you don't grieve these losses you can develop smiling depression.

When you have had bad news, sometimes your normal support structure is also in shock and disbelief. It helps to get assistance from someone outside the family. Family therapy can be helpful.

When to speak with your children. Be sure to give age-appropriate information. What you would say to a 4-year-old is not the same thing you would say to a 14-year-old. My rule of thumb is to always give accurate, current, reliable information, that is age-appropriate. If you don't know what that is, check with someone who understands developmental psychology.

What's the best way to cope with fear? The first thing to do is to get all the information you can. Get educated. Look for the experts; get knowledge. Often that helps a great deal. If the anxiety and anger is daily, then you probably want to consult with a psychology professional.

We do not know that VHL causes these emotional problems. I think we all know that the chemistry in our bodies is very complex. There is a link between stress and physical illness, although it's a weak link. There is growing information that what you think has a lot to do with your compliance with treatment, with your lifestyle, whether you choose to exercise or not, how you think about something does make a difference. And it's a factor to consider. It's not a cause of VHL. But it's just as important to maintain our psychological health as our medical health. Be well.

Update on Clinical Trials

Joyce Graff, Editor

So far there is still no "magic bullet," but we are all searching!

The trials I know about are the following. If you know of another one, please let me know.

(1) Novartis PTK-787, trial headed by Dr. Patrick Wen at Dana Farber Cancer Research Institute in Boston. To qualify, you have to have at least one measurable lesion of the brain, spinal cord, or eye, which cannot be treated by normal treatments without causing you harm. This drug has had good response in the kidney, but this particular trial is designed to measure the effectiveness of PTK with tumors of the central nervous system. This trial is currently OPEN and recruiting patients. Contact Dr. Wen +1-617-632-2166 or one of his nurses Debra Conrad Gigas, RN +1-617-632-6327, Lisa MacDonald, NP +1-617-632-5925

(2) Novartis PTK-787, headed by Dr. Daniel George at Duke University in North Carolina. This trial is NOT CURRENTLY OPEN but is expected to open by about June 2004. This is the same trial as (1) above, at a second site. They are interviewing patients at this time. Contact Dr. Daniel George, +1-919-668-8650

(3) Bayer 43-9006 is doing well in trials with kidney cancer. Patients with brain lesions have been excluded from the kidney cancer trials. There is a proposal to open a study of Bayer 43-9006 with other VHL tumors in Philadelphia and Boston which is wending its way through the forest of approvals. This trial is NOT CURRENTLY OPEN but is expected to open by early 2005. People being seen at the CCC's at University of Pennsylvania (+1 (215) 662-4740) or Mass. General Hospital (+1 617-724-1971) may wish to discuss this possibility with those CCC's.

(4) The Urologic Oncology Division of NIH under Dr. W. Marston Linehan will be starting within the next few months a pre-surgical therapy for kidney cancer. This initial study will be limited to a small number of people with VHL who need kidney surgery. Patients will receive 17AAG from InvivoGen, an analog of geldanamycin, for three months, and then the surgery will help us learn whether the drug is achieving the right response within the tumor. If this phase is successful, the study will be opened more widely. To inquire, contact Dr. Marston Linehan, +1.301.496.6353.

Most of the other kidney cancer trials are currently limited to people with metastatic kidney cancer, as we are still learning safety information about many of these drugs.

(5) Avastin from Genentech is in the category of angiogenesis inhibitors expected to have some usefulness with VHL. It has recently been approved for use with colon cancer, but only in combination with certain chemotherapy drugs. A trial of Avastin and Interferon compared to Interferon alone has opened within CALGB institutions, and trials of Avastin and Interleukin-2 are being discussed but will be limited to patients with metastatic (Stage IV) kidney cancer. See www.calgb.org for participating institutions.

(6) A study at Baylor is identifying targets through tissue staining, looking for VEGF, bFGF, PDGF, C-KIT, and EGFR. Depending on what is positive, several drug combinations are being tried, including Gleevec, Avastin, Ebitux, Revlimid, and thalidomide. Call Dr. Robert J. Amato, Baylor College of Medicine, Houston, Texas, 713-798-1194.

Eye Lesions:

(7) Lucentis (previously named rhuFab) from Genentech, a drug being used for Age-related Macular Degeneration (AMD) is in the process of gaining approval for use with AMD. It is currently being controlled carefully for use ONLY with AMD, in order not to complicate its approval for AMD. Within the next several months, Dr. Emily Chew at NIH expects to open a trial of Lucentis for VHL. This trial is NOT CURRENTLY OPEN, but you are welcome to contact Dr. Chew about the trial. This is an angiogenesis inhibitor, injected directly into the eye, and the study is limited to testing the effectiveness of the drug on eye tumors. Because of the way it is administered, it will likely have little or no effect on other tumors. Inquire with Dr. Emily Chew or her coordinator, Katherine Shimel at +1.301.402.2863

(8) Macugen from EyeTech is also an injectable into the eye, being tested for AMD. Once this is approved for AMD, it is expected to be made available for trial with other conditions. The "wet" form of AMD is caused by leaky blood vessels under the retina, similar in many ways to the process experienced in VHL. See article page 12..

6th International VHL Medical Symposium, May 20-22, 2004

Kochi, Japan, hosted by the Kochi Medical School

see <http://www.6thvhl2004.org/> for details

Thank God for my Family

Alexandra Morais, New Jersey



Alexandra Morais and her son in San Antonio. Her father is behind her shoulder, and husband Jose is behind the camera

I was diagnosed with VHL in the year 2000 after a long clinical history. I recall my first brain surgery back in 1985. I was in my home country of Colombia, South America. I was getting out of college and thinking I had my whole life in front of me. The doctor told me it was a benign tumor. I had surgery to remove the tumor, then physical therapy, and soon after I was dancing again like nothing ever happened.

That same year I came to live permanently in the United States. In December of 1986 I got married and in 1988 my son was born. At the end of my pregnancy I experienced some health problems. I had pre-eclampsia along with some neurological problems. Thank God my son was born healthy.

Ten days after his birth I went back to Colombia for another surgery leaving behind my newborn child and my husband. That was one of the most difficult decisions I ever had to make. I knew I needed to care for my health, and my neurosurgeon in Colombia was then the best option. I stayed there for two months, had surgery, and came back to the U.S.

In 1991 I had an MRI follow-up and what a surprise: I had two tumors in my brain. This time I was referred to a local neurosurgeon. He recommended surgery right away. After that surgery my mobility was no longer the same, but thank God I was still able to live a normal life. The neurosurgeon recommended that I have yearly MRIs to follow up on the progress of the tumors, but there was still no diagnosis. In 1992, after my routine MRI, I had to undergo a surgery in the thoracic spine, and six months later I was back to work.

In the year 2000 I went for a physical and complained to the doctor of a lower back pain. He ordered an ultrasound of the kidneys and after that came a CT scan. Right after that CT scan my primary doctor diagnosed me with VHL, a disease that

even he was not familiar with. He recommended that I go to the internet, and that is how I found out about the VHL Family Alliance.

My primary doctor referred me to a surgeon at Columbia-Presbyterian Hospital in New York City who recommended surgery in both of my kidneys. I had surgery in my left kidney first, as the doctor wanted to make sure that at least one kidney would be working properly in case he needed to remove the other one. Six weeks later I had the second surgery and thank God my kidney was spared. It was explained to me that the tumors in my kidneys were cancerous, but that I did not need any special treatment because the tumors were contained.

Three months later I had brain surgery due to a cyst located in the lower part of my brain, the cerebellum. I mention the location because mostly all my surgeries had involved the cerebellum, which is the balance center. The brain tumors left me with balance problems (ataxia) and I now have more difficulties walking. Due to my poor balance after so many surgeries I now walk with a cane.

After my husband went to the internet and looked in the website for VHL, we finally understood the reason for all my surgeries. Since VHL is a genetic disease I went for a DNA test to confirm the diagnosis. There is no apparent history of VHL in my family. Recently I began the stressful wait for a second opinion, as my kidney surgeon thinks my right kidney needs to be removed.

When I was first diagnosed with VHL, I wished that I had not gotten married and had a child. However, throughout the years I learned to thank The Lord for my husband and my son. I truly believe I have been able to overcome these trials due to their loving support and hard work. Of course I have to mention the support of my

parents, brother and my little nephews (2 and 3 years old), who with their innocence make me forget the difficult times. There is no question in my mind that God had a purpose to put me around people that truly love me. I have learned to appreciate the simple things in life. Even though family has always been important to me, now more than ever I believe with the support of a loving family one is able to overcome any obstacle.

I also want to thank the VHL Family Alliance for the encouragement and support through the newsletters. By reading the newsletters I learned that I am not alone in this battle and that we need to support each other.

A year ago I started to answer the hot line in Spanish as I felt I needed to do something to help people that were in similar conditions as me. So far the response has not been what I anticipated, but I want to take the opportunity to invite the Spanish speaking population to call me at 1-800-767-4845 or to write to info-es@vhl.org. This toll-free number works from the U.S., Canada, and Mexico. I welcome ideas on how to publicize this service to members of the Hispanic population who may be struggling with this illness.

As I finish this entry I want to inform you all that the doctor from the National Cancer Institute contacted me a couple days ago and I was told that currently there is no need to have my right kidney removed. My thanks to VHL Family Alliance for referring me to the NCI.

Finally, I advise all of you to trust God during your hard trials and you will find comfort.

VHL Information Service in Spanish Language

1-800-767-4845

toll-free US, Canada, Mexico

From outside these nations, please call

+ 1.732.699.9090

info-es@vhl.org

Atención: Existe un teléfono gratuito con servicio en habla hispana para EEUU, Canadá y México, el 1-800-767-4845.

También hay una dirección de e-mail a la que puede escribir en español
info-es@vhl.org

La Alianza Familiar VHL proporciona información para mejorar el diagnóstico, tratamiento y calidad de vida de las familias afectadas por la enfermedad de VHL.

Doctors attack 'benefits' of low-carb diets

The reason low-carb dieters often lose weight and sometimes show improvements in their cholesterol, blood sugars, and blood pressures is because they are, in essence, sickened by the diet, writes a doctor in a science journal published in March 2004.

John McDougall, an advisory board member of Physicians Committee for Responsible Medicine (PCRM), explains in his letter to this month's *Mayo Clinic Proceedings* (vol 79, no 3, p431) that low-carb diets can result in a metabolic state called ketosis that also occurs during severe illness, resembling the common side effects of cancer chemotherapy such as fatigue, nausea, and loss of appetite.

People on low-carb diets who become ill enough to experience loss of appetite are taking in less fat and cholesterol, since they are consuming less food overall. It is this same mechanism that results in cholesterol levels falling in cancer patients, he suggests.

The low-carb Atkins diet has seen a strong following in the UK over recent months, with an estimated 3 million people changing to the regime to lose weight. Although some suspect it is merely a fad, the diet has stolen sales from other weight management products in North America, including the leading brand SlimFast.

There is rapid development of "low-carb" brands also in the US, with some of these making recent entries to the UK. However nutritionists and medical experts continue to debate the health aspects of the diet, with many citing evidence to show that high-fat, low-carbohydrate dieters risk clogged arteries, heart attack, colon cancer, and kidney failure. Studies also show that even one fatty meal can increase the risk of a cardiac event immediately following the meal, says Dr McDougall.

"A better approach is to encourage people to eat foods that promote both ideal body weight and health—those from a "high complex carbohydrate, low-fat diet," he said. "For example, people living mostly on high-carbohydrate rice and vegetable dishes in Asia are trim throughout their lives with almost no risk of heart disease, diabetes or our common cancers."

Presymptomatic DNA Testing for Children

At the recent Annual Meeting of the American College of Medical Genetics, Sharon Terry, President and CEO of the Genetic Alliance, participated in a point-counterpoint presentation on the question of offering presymptomatic testing of children for genetic disorders for which there is no treatment. Following 12-minute presentations by both sides, there followed a lively half-hour discussion among the attendees on this delicate topic. The VHL Family Alliance is a member organization of the Genetic Alliance.

Pro position for:

Children may be offered presymptomatic testing for genetic disorders for which there is no treatment at the request of the parent or guardian.

I've agreed to take the pro side of this question. I took it reluctantly, because I felt very middle of the road about this until preparing this argument. In the course of the preparation, I convinced myself!

I put this issue out on the Genetic Alliance MemberForum – the listserv for lay leaders of genetic advocacy groups -- about 4 days ago and it has engendered the best discussion yet! So my comments are infused with the passion of my colleagues.

Like all ethical questions, we like to pretend to ourselves that it is black and white. But the shades of grey are especially evident when one acts as if they are simple choices: personal choice and societal good; disease and health; treatment and management; autonomy and authority; benefit and risk; advantage and disadvantage. But in fact they are points along a continuum of shades of grey.

I begin by sharing with you a message from one of our members. I've changed the names to protect their confidentiality. Sally writes:

"Our son, Michael, was diagnosed 13 years ago, at the age of 12, with Niemann-Pick Disease Type C. NPC is a rare, neurodegenerative, life-limiting, metabolic disease for which there is no cure and, as yet, no effective treatment.

"When Michael was diagnosed we were told that the disease was autosomal recessive and that there was a one in four chance that his older brother, Rick, aged 14, might also have NPC. We were asked if we wanted to have Rick tested - we said, "No". Rick was physically very fit and, as far as we could tell, did not seem to have any obvious symptoms of the disease. We felt that we had enough to cope with dealing with Michael's diagnosis and the severe seizures that he was having at the time.

"After a while however, we did begin to wonder if perhaps the recent difficulties that Rick was having in some of his subjects at school might possibly mean that he too might be affected. (There were no support groups for NPC at that time and we did not know of any other families we could speak to.) Eventually we decided that we really needed to know, to set our minds at rest and free our energies to taking care of Michael, or to gather our strength to deal with the emotions and

practicalities of the deterioration and death of both of our sons. We arranged for Rick to be tested and waited three months for the results of the skin biopsy.

"We were devastated when we learned that he too had NPC.

"Michael died six years after his diagnosis at the age of 18 years.

"Rick has had a much slower progression of this disease. He is now 27 years old, still physically strong but he struggles with dementia and some mobility difficulties.

"A year or two after Rick's diagnosis, when he was still apparently well, I asked him if he ever wished that he did not know that he had NPC, that he had never been tested. He said, "Oh no, Mum, now I know I am not stupid. I know there is a reason for some of the things I can't do". When we had thought that he had no obvious symptoms, Rick had been struggling to understand why he was not able to keep up with his peers, why there were some things that he could not do as well as he wanted to.

"In spite of the horror of the diagnosis, I am glad that we decided to have the test, so is Rick. He has been able to make life choices that he might not have made, had he not known. Knowing that he has NPC has enabled us to support him in his choices and not put undue pressure on him in other ways.

"Rick is very aware of the effects of NPC - he has seen what it did to his brother and to many friends he has made over the years. He has been very involved in meeting other families and in taking part in research. He is currently taking part in a clinical trial of a possible new therapy that might slow down the progression of the disease. He remains positive that an effective treatment will be found. Rick is a wonderful, very determined, young man who happens to have NPC and, we are told, is an inspiration to many other families living with this disease.

"Are we in favor of testing presymptomatic siblings? Definitely yes!"

Sally has said beautifully a number of things that give a face to this argument.

The American Academy of Pediatrics says, "little direct benefit accrues to a child simply from knowledge of his or her genetic status." The American Society of Human Genetics and the American College of Medical Genetics, have joined together under this policy: "children may not accrue benefit

from being tested for genetic disorders.” They do urge caution before ‘dismissal’ of parent requests.

These policies are based on benefits and harms – the desire to do good, and not to do harm. But what these organizations have determined as policy doesn’t quite jive with Sally’s perspective.

What are the benefits? When I asked my colleagues, they cited these as the benefits: preparation – for lifestyle (for the family and the child), choosing caregivers and specialists, financial planning, choice of job, educational choice, finding a support group, securing insurance.

Parents felt that they could benefit from cultivating relationships, watching for signs and symptoms of the disease, alleviating their anxiety, building their support structures, promoting research and building registries, and beginning the long road to acceptance.

When any one of us who lives with disease looks at this choice, we see it very differently than those who do not live on this side of the line. The biggest question for us is: who determines what is health and what is disease, what is whole and what is not? Variations in health are normal.

We do not accept that we are speaking about something for which there is no treatment – again, we feel that treatment is defined rather narrowly by the medical community. Person after person on our listserve gave examples of treatments that might not be considered treatments – these were: a different lifestyle, changes in schooling, special diets, preventative measures. Even in the absence of medical therapies, many parents felt there was still a great deal of ‘management’ to undertake. They did not ascribe to “What will be, will be,” but thought that, particularly if they could build a group of affected families, they could encourage research. There was a sense of urgency – we need to start now beginning to accept others and accept disease – and by doing so, we will create an environment for true public health maintenance.

Parents do not want the doctor making decisions for them, they want to make the decisions for themselves and their children. Some people argue that the child should be free to discover their affected or unaffected status as adults. This argument is made by healthy people – it doesn’t make sense to those of us on this side of the line. These healthy people believe that if a disease is revealed, then the child will be devastated. This is not our experience and it is not the experience of children such as Rick. Living with disease or wondering if you have disease can be harmful or beneficial, depending on the circumstances.

In addition to medical arguments, there are a number of psychological arguments:

Behavior – it is often said that the behavior of these children will be worse for the testing. Several



Making VHL connections at the Tampa meeting.
See page 16 for a meeting near you.

studies by Michie (*J. Med. Gen* 1996) and Järvinen (*Pediatrics* 2000) showed that children in these situations did not have behavior that was worse than their peers.

Stigmatization – Jolly, in his article *The Genetic Testing of Children* refutes this. McConkee-Rosell says that only 1 in 28 women carriers of fragile X felt it would be worse to know as a child. All the rest were not worried about stigmatization.

Fanos, in a study on Ataxia Telangelectasia, determined that testing is well tolerated provided there are the proper supports.

John Twomey, author of a paper: *Genetic Testing of Children: Confluence or Collision Between Parents and Professionals?*, an article in the *American Journal of Nursing*, determines that when parents make decisions within the family framework, the parents are placing a higher value on the child, nurturing him or her in the present, not some future unknown to be preserved with all options open. Parents comfort their children, help them prepare for future decision-making and include them in the family to help make them part of the family fabric.

He suggests that families are not using medical principles, nor are they measuring things by the psychological metrics, but instead are using ‘ethics of care’ as a measure. In an ethics of care environment, rules are suspended in favor of strengthening relationships. The work of Sherwin suggests that the family doesn’t seek universal guidelines, but instead seeks its own balance in a dynamic way.

Thus parents fill a roll that no professional can ever fill, nor can a professional ever make a decision for a family.

Twomey goes so far as to say: “No universal policies should be [legislated] by any bodies that recommend absolute limits on genetic testing of children under any circumstances. The best way to



*Gale Lugo, Florida
Chairman, speaking
at the Tampa
meeting*

protect the interests of the child is not found in protections from theoretical harm, but rather in means for educating all involved in the decision making process."

My two children have a late onset genetic condition. They've lived with this knowledge for nine years and two months. It will cause blindness and a host of other difficulties. They have seen hundreds of people blind and disabled from pseudoxanthoma elasticum (PXE) because of traveling the world with us as we built the extended family we call PXE International.

A few months ago, Elizabeth began a small tirade at dinner – she is now 16, and her brother Ian, age 14, chimed right in with her. They very heatedly told us

that their disease was not a disease – it was a gift.

They said that they would choose no other path – that having PXE makes them no less a person now that they are aware of it, that our perceptions of health are in the box, and that they, and all people with genetic conditions, have a right to live in this life the way they will – even if others think it is less than whole and not complete. They said they would choose to know about the risks they face because these nine years of knowing have gifted them with more depth and purpose in life than they think they would have gotten in any other way. They agree with Rick.

One of my wise peers points out: *"It is not the cards you are dealt in life that counts, but rather how you play them."* You cannot maximize your playing of the cards if you don't know what they all are. I choose to know for my children, that they might be integrated into the fabric of our life. I want us to have the time and opportunity to build the support, to create structures and actions upon which to hang hope, and at the end of the day, to know, and in the knowing experience less harm and less risk, all the while preserving our autonomous decision making, nurtured in the loving environment of our own family dynamics.

I think the decision to test should be the parents' decision, and the health professional community should offer the support and expertise necessary to make that experience a healthy and whole one.

Thank you.

Preventing Infections

Who is at risk?

- People who have had major surgery within the last 6-12 months.
- People who have had radiation treatments, including stereotactic radiation within the last 12-24 months.
- People who have a transplant of any kind
- People whose systems are weakened by chronic illness, asthma, diabetes, or use of steroids
- People being treated with chemotherapy

People who have more than one of these risk factors should consider themselves to be at higher risk and be even more scrupulous.

Prevention

Most of the keys to preventing infection are those common-sense things we learned in kindergarten, with a few extra precautions.

- Wash your hands frequently with soap and hot water, especially after shaking hands or playing with children, and always before eating.
- Use gloves to prevent contact with fecal matter, especially when changing cat litter, bird feeders, and

baby diapers. Wash hands thoroughly with soapy water after any such contact.

- Keep hands away from the eyes, nose, and mouth to avoid carrying germs to your mucous membranes.

- Be extra careful to wash hands and sterilize contact lenses. People at elevated risk should consider discontinuing use of contact lenses until their systems are stronger.

- Gardening may not be the best activity for people at risk. Many fungi live in the dirt, and soil disruption of any kind can lead to an increased risk of infection. Fungal spores travel through the air, and dry or dusty dirt can cause the spores to spread. Avoid construction or renovation sites.

Any outdoor injury to the eye should be considered very serious, and should be seen immediately by a physician. Any infection should be considered serious, especially infections of the sinus or lungs.

If you have a fever higher than 100.5° F. (38° C.) or if you have cold symptoms for more than 3-4 days, you should see your doctor.

Our thanks to Andrea Snyder, M.P.H., Centers for Disease Control, Atlanta, Georgia, for her assistance in preparing this article.

LoriAnn's Gift

by Janice J., New York

LoriAnn Schindel Marquardt lived a very full life. She refused to let her disease and disability define her life. According to one teacher she was the student who brought life and vitality to the room. Life was exciting and was to be explored.

We lost LoriAnn in December -- not to VHL, but to a fungal infection. We wanted to share her story with you both to celebrate her life, and to help others avoid the devastating effects of infection.

LoriAnn loved music and dance and would dance to the TV before she could walk. She became an outstanding flautist, as a soloist and as a member of the Minnesota high school All-State Band. Her vocal accomplishments took her to Europe with a select group in her sophomore year. Her sense of music caused her flute teacher to comment, "LoriAnn sings through her flute."

LoriAnn was born in Jamestown, North Dakota. She grew up in Duluth, Minnesota. She graduated from Bethel College in St Paul, Minnesota, with a degree in English Literature. She and Larry Marquardt were married Oct 26, 1991. They lived in Richfield, Minnesota. Besides her husband she is missed by her parents Bonnie and Elmer Schindel, her brother Jay and his son Jacob, her step children Michael and Gretchen, and grandson Jackson.

LoriAnn's journey with VHL began before she could remember, as her grandfather battled the disease. When she was only a year old her mother had her first brain tumor. At age 15 LoriAnn herself was diagnosed and treated for lesions on the retina. She had numerous treatments in her eyes and eventually lost her right eye. Three retinal detachments in the left eye finally left her legally blind. Her blindness did not deter her from pursuing her career as an Administrative Assistant at United Resources Network (URN), a division of United Health Group, that coordinates the location and distribution of human organs for transplantation.

At the age of 22 LoriAnn lost one and one-half kidneys to kidney cancer. The half kidney that remained served her for five years, until it too needed to be removed. Still she maintained her work at URN. After a full day at work she spent three evenings a week on dialysis, and was ready to return to work in the morning. She continued to travel and be involved in many activities, always planning ahead so she was near a dialysis center at the right time.

Her father had wanted to give one of his kidneys, but he came down with cancer that same year. His brother, LoriAnn's uncle from British Columbia, gave LoriAnn her first new kidney. She went into

LoriAnn
Schindel
Marquardt
2003



rejection, the kidney failed, and she returned to dialysis. In March 1999 she received a cadaver kidney which served her the rest of her life.

She had five brain tumors treated with stereotactic radiation. Throughout her series of challenges, she attacked life with vigor. She coped with her own disability and helped share her "can do" spirit with others. She helped facilitate a support group at Vision Loss Resources and became an advocate for people who needed help negotiating the medical system. She took up cooking as a hobby. She looked forward to preparing a meal for friends or family, sometimes from others recipes and sometimes her own creations. In spite of her health issues People who first met her could not tell that her health was compromised as she did not dwell on what she did not have, or could not do, but was busy living each day as best she could.

The VHL issue that sneaked up on her was her pancreas. She was taking pills for enzyme replacement and knew about the risks of low blood sugar, but has not been educated about the risks of high blood sugars. Organ transplant patients are subject to fungal infection. There is no cure for many fungal infections, especially if not treated quickly. Bacteria, viruses, and fungi are present all around us at all times. A healthy body has the immune system to protect us. High blood sugar lowers the immune system, along with other risk factors. LoriAnn's blood sugars had spiked. Because of her generally poor health she did not associate this feeling with a new problem. She lost her energy and lost weight, but only in retrospect did it become clear that her blood sugars had been out of control for some time.

Those at risk for fungal infections should take every precaution possible and should treat every infection as a possible threat to their life. They should see their doctor immediately and discuss the possibility of a fungus, as it is generally not the first issue that a physician will consider.

When asked what she saw ahead of her in death, she said, "God is in charge and everything is OK."

She died just as she lived her life,

With faith shown at its best.

Age-related Macular Degeneration (AMD)

by: Altheada Johnson, MS, RD

AMD is not a feature of VHL. Nonetheless, the vascular irregularities in the macula are often treated with laser as in VHL, and angiogenesis inhibitors are being developed to slow the advancement of vascular damage in the retina. These same drugs are expected to be helpful with VHL. Altheada provides us with a discussion of AMD to help us understand the similarities between AMD and VHL, and suggesting some nutritional supplements that have been found to be helpful with AMD.

What is a macula? The macula is the small, yellowish central portion of the retina and it is the area providing the clearest, most distinct vision. When one looks directly at something, the light from that object forms an image on one's macula. A healthy macula ordinarily is capable of achieving at least 20/20 ("normal") vision, even if this is with a correction with glasses or contact lenses.

What is age-related macular degeneration? Macular degeneration is a hereditary ocular disease. Age-related macular degeneration (AMD) is not hereditary, but may sometimes be a consequence of aging, and is the leading cause of irreversible blindness among Americans 65 and older. "Dry" macular degeneration generally is caused by a thinning of the macula's layers, and vision loss typically is gradual. However, tiny, fragile blood vessels can develop underneath the macula. "Wet" macular degeneration can result when these blood vessels hemorrhage, and blood and other fluid further can destroy macular tissue, even causing scarring. In this case, vision loss can be rapid, over months or even weeks.

Macular tissue destroyed by either dry or wet macular degeneration cannot be repaired. In the case of the wet form, a special laser can be used to seal the leaking blood vessels in the retina. However, (1) the tiny spots where the laser burns the retina will lose vision permanently, and (2) other blood vessels may leak in the future, requiring further laser treatment.

The earliest symptom of macular degeneration usually is persistently blurred vision. As more cells of the macula are destroyed, objects become distorted (for instance, straight lines become crooked). Eventually, a small blind spot in the central visual field can develop and grow in size. This can progress to the point of "doughnut" vision, where people's faces are unrecognizable when looking directly at them, yet peripheral vision remains unaffected.

How is all of this related to VHL? There is no direct connection between VHL and AMD. Sometimes, people with VHL have macular degeneration,

glaucoma and/or cataracts. In addition, treating the angiomas on the retina, resulting from VHL, can cause conditions similar to AMD. But the primary reason for this article is to help you understand why a treatment for AMD might also be effective for VHL. Angiomas are small knots of blood vessels that can, themselves, leak and damage the vision very much like some forms of macular degeneration.

Naturally occurring carotenoids in the macula, *lutein* and *zeaxanthin* (molecular cousins of beta carotene and vitamin A), have been shown to be effective protectants against degeneration of the macula. These pigments absorb and filter out near-to-blue ultraviolet radiation, acting essentially as built-in macular "sunglasses" -- which potentially is the most damaging electromagnetic radiation reaching the macula. The greater the amount of macular pigment, the less the risk of macular degeneration. Lutein and zeaxanthin are found particularly in yellow fruits and in green leafy vegetables (especially vegetables such as spinach, kale, collard greens, and broccoli), and in eggs, and as nutritional supplements. A half cup a day of one of these vegetables (cooked) will provide the recommended daily amount.

Getting these vitamins from food rather than pills provides other phytochemicals that might act in a synergistic way, to help the absorption and utilization of the nutrient of interest or to aid in the way it protects the body from damage.

Treatments which are effective at reducing the fluid leakage beneath the retina, or in controlling the proliferation of blood vessels in the retina, might also be effective in the treatment of the fluid leakage and blood vessel proliferation that occurs in VHL. Clinical trials of these agents are beginning to open this year.

For the carotenoid levels of foods, and other nutritional hints, see www.vhl.org/nutrition

Targeting VEGF

There are several competing new drugs designed to stop macular degeneration before the scar forms. Called anti-neovasculars, they take aim at the body's prime stimulator of blood vessel growth, a protein called Vascular Endothelial Growth Factor.

VEGF is also one of the primary targets in dealing with VHL tumors.

Ordinarily, VEGF is a healer, repairing injured tissue throughout the body. In wet macular degeneration VEGF goes haywire, triggering growth of the oozing blood vessels.

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VHL Family Forum

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 Peggy Marshall, by Katie McConnon
 Melissa Minster an the Minster Family, by Ronald Pelton, Albert Young
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 Debra Dankenbring Reed, by John & Mary Meehan, Third National Bank of Sedalia
 60th Wedding Anniversary, by Joyce Reed
 Kelly Roberts, by Norma & Dick Maresco
 Philip & Michel Roche, by Tamara Phillips
 Roche Family, by Stacey Brooks
 Lisa Rossman, by Marcia & Charles Rossman
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 The Schneiders, by Rick and MariBeth Walker-Schneider
 Rev. & Mrs. John Sewell, by June & Harry Wilcox
 Ruby Wedding anniversary of Michael & Frances Smith, by Mrs. Frances Smith
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Editor: Joyce Wilcox Graff, 1-617-277-5667 (day), 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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Meetings! Come and form some VHL Connections!

Charlotte, North Carolina, April 17

Community Conference Room, Presbyterian Hospital Matthews, 1500 Matthews Township Parkway, Matthews, North Carolina 28106-3310

\$25 per person in advance, \$30 on-site. Registration includes lunch.

Presentations will include: Advances in neuro-surgical treatment with Dr. Stephen Tatter of Wake Forest, and Information about the opening of a second site at Duke University of the trial of Novartis PTK-787 to determine its effectiveness with the brain, spinal, and eye tumors of VHL. Our host, Darlene Byrd, will introduce the patient and family services of The Health Library.

Register online or on page 15. For additional information, call Audrey Clifton at 910-867-0352.

Dallas, Texas, April 24

University of Texas Southwestern Medical Center, Simmons Biomedical Research Building, Room: NB 2.402/403, 6000 Harry Hines Blvd.

Dallas, Texas 75390, 11:30 to 5:00

No charge, light snacks will be provided

Presentations will include:

- Bruce Mickey, M.D., Neurosurgery
- Arthur Sagalowsky, M.D., Urologic Oncology
- Rand Spencer, M.D., Texas Retina Associates
- Linda Robinson, M.S., Genetic Counselor

For additional information, contact Michelle Sanchez, 281-403-0109, or Clenton "Fin" Winford II, 972-264-6886, us-tx@vhl.org

Kochi, Japan, Medical Symposium

see www.6thvhl2004.org for details

Join us in New York

at the VHLFA

Annual Membership Meeting

Saturday, June 19th, 2004, 9 AM-5PM

Crowne Plaza/La Guardia Airport

104-04 Ditmars Blvd., East Elmhurst, NY 11369

\$99/night 718-457-6300

All attendees should make their own hotel reservations

Conference Registration

can be completed at www.vhl.org/meetings or see page 15

Presenters include:

- Dr. Gladys Glenn from the NIH. Dr. Glenn will speak on VHL screening.
- Dr. Jay Klancnik, from Manhattan Eye, Ear & Throat will discuss eye issues in VHL.
- Dr. Wendy Chung, Geneticist, Columbia-Presbyterian Medical Center, on DNA testing
- Dr. Debra Shabas will speak about care-giving.

The Program will also include **VHL Connections** - an opportunity for us to share our experiences.

Conference Registration is \$25 per VHL patient or family member and \$50 for genetic counselors, nurses, doctors, social workers and other health professionals.

CEUs will be available for genetic counselors.

Optional tickets for Beauty and the Beast available at the group rate of \$35/seat for Saturday night.



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