

Von Hippel-Lindau Disease: An Overview

Michele Inglese

Von Hippel-Lindau Disease is a rare, familial disease involving the presence of multiple tumors, angiomas, and/or hemangioblastomas. Cysts are found around the tumors. The kidneys may not be the primary sites of involvement. The primary sites may be the eyes, the brain, the adrenal gland, the pancreas, the liver, spinal cord, and/or the kidneys. The tumors are named depending on the site. Tumors involving a nest of blood vessels are termed angiomas or hemangioblastomas. Hemangioblastomas can occur in the retina, brain, or spinal cord in patients with VHL. These hemangioblastomas are usually not cancerous. When the eyes or brain are affected, there are multiple tumors. Renal lesions may be cystic, combined cystic-solid, and solid renal cell carcinomas. In early studies, renal cysts were found to be present in about 60% of patients with VHL, but new screening technologies are finding the incidence to be higher (Choyke et al., 1992). If the kidneys, pancreas, or adrenal glands are affected, there may be a single tumor at the onset or there may be multiple tumors. The kidney tumor may be or is likely to become cancerous (NINDS, 2005). The tumors spread and make this disease a multi-system disease.

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Von Hippel-Lindau disease is a rare, familial disease consisting of multiple tumors, which can present in the eyes, brain, adrenal gland, pancreas, liver, spinal cord, kidneys, or other areas of the body. The renal tumors are primarily renal cell carcinoma. Renal involvement may be the primary manifestation, but most frequently it is a secondary manifestation. This article presents an overview of von Hippel-Lindau disease and the nursing care of the patient with renal involvement.

Goal

To inform nephrology nurses about the manifestations and treatment for the renal involvement of Von Hippel-Lindau Disease

Objectives

1. Describe the genetic basis for the tumor development in Von Hippel-Lindau Disease.
2. Compare the treatments used for patients with renal carcinomas caused by Von Hippel-Lindau Disease.
3. Develop an education plan for patients with a family history of Von Hippel-Lindau Disease.

Eugen von Hippel, a German ophthalmologist, first described the retinal tumors in 1904 (von Hippel, 1904). Arvid Lindau, a Swedish pathologist, made the association between the brain, kidney, and epididymis tumors as well as the familial connection in 1926 (Lindau, 1926). The gene that causes VHL was identified in 1993 and is located on chromosome 3, more specifically chromosome 3p25 (Kaelin, 2004). This malfunctioning tumor suppresser gene allows the tumor to reproduce and spread. This disease is passed on as an autosomal dominant trait and

usually manifests when people are in their twenties. Males and females are equally affected. Currently, there is no association of risk factors with any specific ethnic or cultural group.

Signs And Symptoms

The signs and symptoms depend on the sites of the tumors. Brain tumors may manifest as difficulty with balance, ambulation, headaches, weakness, etc. Eye tumors may manifest as blurred vision. Kidney involvement may first present as back pain and or hematuria.

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Table 1
Von Hippel-Lindau Disease Resources

American Brain Tumor Association

<http://www.abta.org>

CancerNet

<http://www.cancernet.nci.nih.gov/cancertopics>

Genetic Alliance

<http://www.geneticalliance.org>

National Cancer Institute

<http://web.ncifcrf.gov/research/kidney/vonhip.html>

National Institute of Health NIH Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=vonhippellindausyndrome/show/Educational+resources>

National Institute of Neurological Disorders and Stroke:

NINDS Von Hippel-Lindau Disease (VHL) Information Page
http://www.ninds.nih.gov/disorders/von_hippel_lindau/von_hippel_lindau.htm

National Organization for Rare Disorders (NORD)

<http://www.rarediseases.org>

von Hippel Lindau Family Alliance

<http://www.vhl.org>

Diagnosis

The diagnosis can be made by family history and with the presence of at least one of the two following findings: brain or eye tumors, or a single brain or eye tumor along with tumor(s) in the other suspect sites such as adrenal gland, liver, pancreas, or kidney. When there is not a known family history, the diagnosis can be made by the presence of both of the previous findings (Maher et al., 1991).

Renal cysts and tumors are sometimes identified coincidentally during diagnostic tests for other diseases. Multiple cysts found in the kidney may be indicative of other diseases, such as polycystic kidney disease (PKD), and not necessarily VHL. The practitioner needs to differentiate VHL from other cystic kidney diseases. The renal tumors are frequently renal cell carcinoma.

Because VHL is a hereditary disease, all people with known family histories of VHL should be offered the option to be screened using genet-

ic testing to determine if they are affected. The National Cancer Institute has a program called the Familial Kidney Tumor Program, which follows patients with VHL (NCI, 1996).

Treatment

The treatment of VHL depends on the sites of involvement, severity of the symptoms, and whether the tumors are benign or malignant. Treatment may include conservative management, radiation, or various levels of surgery. While Neumann and Zbar (1997) say that surgery "is the only accepted treatment of renal cell carcinoma" (p. 21), others advise a more conservative approach.

Surgery may include the complete removal of one or both kidneys, a radical nephrectomy or what has been termed nephron-sparing surgery (NSS). When both kidneys are removed, renal replacement therapy must be initiated. In NSS, the tumors are removed and the procedure may

be repeated. The desired result of NSS is to save renal function. Steinbach et al. (1995) found NSS to be an effective initial treatment, but noted that the patients needed to be followed closely as they may have locally recurrent renal cell carcinoma. Hes et al. (1999) described a comparison of the two types of surgery with comparable renal cell carcinoma growth results for each procedure and preservation of renal function. Roupret and colleagues (2003) studied NSS using a larger sample size and found NSS effective in treating renal cell carcinoma and preserving renal function. Other newer methods of treatment, including ablation, are being used to treat smaller tumors.

Individuals with VHL need to be monitored at intervals to determine progression of the disease. Monitoring includes current history and physical, eye examinations, and diagnostic tests such as computed tomography (CT).

Prognosis

The prognosis depends on the sites, involvement, presence of cancer, and metastasis among other related factors. Renal cell carcinoma is more likely to occur as one ages and has a guarded prognosis. Mortality is associated with brain tumors and renal cell carcinoma.

Nursing Care

The nursing care of the person with VHL includes a thorough assessment with a focus on the patient's knowledge of the disease, history, and current manifestations. The patient who is newly diagnosed and who has no known family history of VHL will differ from the patient with a known family history who has prior knowledge of VHL.

For the patient with a family history, the nurse should assess the patient's knowledge of VHL and identify any misconceptions, if present. As this disease is so rare, it is possible and likely that the patient with a family history and in-depth knowledge base of VHL may have more information about the disease

Table 2
Possible Nursing Management of Patients with VHL and Renal Cell Carcinoma

Situation	Nursing Diagnoses	Nursing Outcomes The patient will be/demonstrate:	Nursing Interventions
Newly diagnosed with family history	Acute pain R/T* tumors/cysts	Pain-free or satisfactory pain relief	Pain management Medication management
	Fear R/T loss of body function, treatment or terminal illness	Effective coping	Support and comfort measures Relaxation therapy Medication management Collaborative care: counseling
Newly diagnosed without family history	Knowledge deficit R/T	Increased knowledge	Patient teaching
	Anticipatory grieving R/T loss of body function or death	Effective coping Expression of grief	Emotional Support Family Support Grief facilitation
Renal cell carcinoma treated with surgery	Risk for infection R/T surgery	Infection free	Wound care
	Fluid volume excess R/T possible renal impairment	Edema free Euvolemic	Monitor fluids and electrolytes Fluid management
Renal cell carcinoma treated with chemotherapy/radiation	Disturbed body image R/T effects of treatment (i.e., hair loss)	Increased or acceptable self esteem	Counseling Support groups Grief facilitation
	Fatigue R/T anemia	Maximum level of function Activity tolerance	Medication management Patient teaching
	Impaired urinary elimination R/T: Situational retention or renal impairment	Urinary Continence	Toileting schedule Collaborative: urinary catheterization or dialysis
	Impaired tissue integrity	Tissue integrity of skin and mucous membranes Pressure ulcer free	Skin monitoring and care Positioning Oral hygiene and care
	Impaired nutritional status-less than body requirements R/T cancer (insufficient caloric intake, difficulty eating)	Minimal weight loss Acceptable caloric intake	Collaborative: dietary counseling Patient teaching Daily weights Nutrition management

Note: These diagnoses are not limited to the situations listed and may belong in more than one field. For example, knowledge deficit or potential for infection may be appropriate in any of these situations.

* R/T = related to.

than the nurse. It is important to ascertain the patient's perception of the disease and the affected family members' histories to assist the patient in dealing realistically with the diagnosis, care, and prognosis. If the patient has a known family history, it is beneficial for the nurse to know the family members' experiences and outcomes with VHL. If the patient's

experience was observing a family member with VHL who had multi-system failure resulting in death, the patient's response and coping may be pessimistic. Conversely, if the patient observed a family member with VHL who lived for many years, the expectations may be inappropriately optimistic.

For the newly diagnosed patient

without a known family history, the patient and family will need to be introduced to the concept of genetic counseling and the resources available. A list of resources can be found in Table 1. The nursing role is to provide information and support for the patient's decisions. Factors affecting the decisions include the patient's age, understanding, cognitive ability,

cultural practices, family role and influences, coping skills, desired outcomes, and comorbidities (Masny, 2004). The history should include a cancer history for three generations from both sides of the family (Stern, 2004). As the gene has only been identified within one generation, previous generations would not have been confirmed as VHL. Patients and health care providers may only find that there is a strong family history of renal cancer only.

The clinical manifestations will determine the additional nursing diagnoses and interventions. Monitoring of renal function and onset or progression of other manifestations is an ongoing nursing action. VHL is frequently a multi-system entity and manifestations may vary. With the presence of cysts and tumors, pain assessment and management should be included. Possible nursing diagnoses include risk for infection, fear and anxiety related to the diagnosis, acute pain, impaired urinary elimination and (possibly) knowledge deficit (Carpenito-Moyet, 2006). If renal cancer is diagnosed, the nursing diagnoses may include anticipatory grieving. Additional nursing diagnoses would depend on the treatment of the cancer: nephron-sparing surgery (acute pain, imbalanced fluid volume) or chemotherapy/ radiation (impaired skin integrity, imbalanced nutrition less than body requirements, fatigue). These are not inclusive but may vary widely depending on the patient, site, and other influencing factors (see Table 2).

The interventions, outcomes, and evaluations flow from the nursing diagnoses, but consistently these patients would need to be supported through this potentially long-term disease process. For those with poor prognosis related to advanced cancer or brain tumors, the support shifts to dealing with end-of-life issues.

Summary

VHL is a relatively rare disease, however, the potential for renal manifestations necessitate that nephrology

nurses be knowledgeable about the disease. The complex, multisystem nature of VHL challenges nephrology nurses to use a wide range of skills to provide care to a small, but needful, population of patients.

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1. What would be different in your practice if you applied what you have learned from this activity?
- _____
- _____
- _____
- _____
- _____
- _____

GOAL To inform Nephrology Nurses about the manifestations and treatment for the renal involvement of Von Hippel-Lindau Disease.

New Posttest Format
 Please note that this continuing education activity does not contain multiple-choice questions. We have introduced a new type of posttest that substitutes the multiple-choice questions with an open-ended question. Simply answer the open-ended question(s) directly above the evaluation portion of the Answer/Evaluation Form and return the form, with payment, to the National Office as usual.

Evaluation

2. By completing this offering, I was able to meet the stated objectives
- a. Describe the genetic basis for the tumor development in Von Hippel-Lindau Disease
- b. Compare the treatments used for patients with renal carcinomas caused by Von Hippel-Lindau Disease.
- c. Develop an education plan for patients with a family history of Von Hippel-Lindau Disease.
3. The content was current and relevant.
4. This was an effective method to learn this content.
5. Time required to complete reading assignment: _____ minutes.

	Strongly disagree				Strongly agree
	1	2	3	4	5
	1	2	3	4	5
	1	2	3	4	5
	1	2	3	4	5

I verify that I have completed this activity _____ (Signature)