International Journal of Advances in Medicine
Avva VS et al. Int J Adv Med. 2025 Sep;12(5):516-518
http://www.ijmedicine.com
pISSN 2349-3925 | eISSN 2349-3933
DOI: https://dx.doi.org/10.18203/2349-3933.ijam20252542

Case Report

Von Hippel-Lindau syndrome: a rare case report from a tertiary care hospital

Veena Santoshi Avva*, D. Nageshwar Reddy

Department of Medical Gastroenterology, Asian Institute of Gastroenterology Hospital, Gachibowli, Hyderabad, Telangana, India

Received: 09 July 2025
Accepted: 13 August 2025

***Correspondence:**
Dr. Veena Santoshi Avva,
E-mail: veenasantoshi89@gmail.com

ABSTRACT

Von Hippel-Lindau (VHL) is a diagnostic challenge due to the fact that it is rare and as stated above, presentation is also not typical in all cases. It is quite rare as prevalence ranges from 1 per every 30,000-50,000 population per year. Present case is unique in the sense of difficulty in the diagnosis at previous clinics. She had retinal angioma, left renal mass, infertility. Thus, multiple presentations make the diagnosis difficulty and thereby treatment. Hence, it is necessary to present all possible cases which come across during the routine clinical care so that the physicians and surgeons bear this differential in mind.

Keywords: Retinal Angioma, Infertility, Case report

INTRODUCTION

Von Hippel-Lindau (VHL) syndrome affects multiple organs. It is autosomal dominant disease. It is characterised by several tumors or cysts which can be malignant or benign. Hemangioblastoma is the most common tumor seen in VHL. It is benign in nature. They are commonly seen in brain, retina. There can be loss of vision and ataxia. Another common presentation is occurrence of cysts commonly seen in pancreas, kidneys and genital tract.

\Other important most common tumors are pancreatic neuroendocrine tumor and the renal cell carcinoma (RCC).[1,2] It has been estimated that the prevalence of VHL ranges from 1 per every 30,000-50,000 population per year. On an average VHL can be seen at around 18-30 years of age. But it can be seen in infants and elderly up to 70 years of age. There is no gender predilection. The incidence of renal cysts in VHL is 59-63%, that of RCC is 25-45%, CNS hemangioblastoma incidence is 13-72%.[3] The cause of VHL is occurrence of mutation of a gene that suppresses the VHL tumor. This gene is located on chromosome.[3] Due to this, there is production of abnormal VHL protein that leads to degradation of hypoxia-inducible factor (HIF) resulting in uncontrolled HIF upregulation and also the growth factors. These sequence of events leads to formation of multiple cysts and tumors, which are the characteristics hallmark of VHL.[3] The patient presentation depends upon the site at which the cyst or tumor is located.

There can be headache, ataxia etc. in cases of hemangioblastoma of central nervous system. Same tumor in retinal can lead to ocular symptoms even sometimes loss of vision. The symptoms of pheochromocytoma may be vague. Diagnosis is usually confirmed by laboratory and radiologic investigations. Treatment/management also depends upon the cyst or tumor location and its size.[4]

Thus, VHL is a diagnostic challenge due to the fact that it is rare and as stated above, presentation is also not typical in all cases. Hence, it is necessary to present all possible cases which come across during the routine clinical care so that the physicians and surgeons bear this differential in mind.

CASE REPORT

Authors present a case of a female patient who was 27 years of age.

Past history

She presented with pain abdomen intermittently in epigastric region radiating to back at some other clinic. There she was diagnosed as Pancreatitis and was given CREON (pancrelipase) 25,000. But there was no relief of her symptoms. Later she presented to us with similar complaints. Authors carried out further evaluation and management of her symptoms. She gave history of infertility for which she underwent fertility treatments in the year 2022-2023.

She conceived meanwhile and delivered a baby by lower segment cesarean section. During this period, she had left sided pain abdomen which was located in the hypochondriac region and extended till flank. Ultrasonography was done and a left renal mass was noted. For confirmation she was asked to undergo magnetic resonance imaging (MRI) abdomen which was normal. Hence, she was given conservative management with analgesics. She was relieved. But in September 2023, she had similar kind of pain. This time, she underwent CT abdomen which revealed Left renal mass suspicious of neoplastic lesion T1b, Nx, Mx and pancreatic cysts. She underwent partial nephrectomy on 10/10/2023. Histopathology examination was done which revealed that it was clear cell carcinoma.

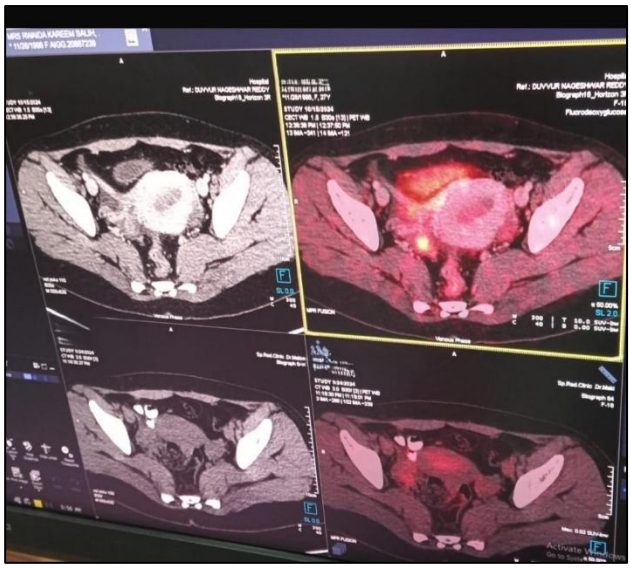

Figure 1: PET CT highlights the cystic lesion of pancreas.

Present scenario

She presented to the institute for further evaluation of her previous problem and evaluation of her pain abdomen. In view of her past history of infertility, multiple pancreatic cysts, episode of pancreatitis and clear cell carcinoma, we suspected Von Hippel Lindau Syndrome. We further evaluated with ophthalmologist and PET CT scan was done. Ophthalmology examination revealed a suspicion of left retinal Angioma. PET CT revealed multiple pancreatic cysts, renal cysts and adrenal cysts (Figure 1). We sent the sample for VHL analysis which was heterozygous positive and confirmed diagnosis of VHL. Authors advised her pancreatic enzyme supplements of lower dose in view of pain. The patient responded to this conservative management. We advised for routine check-up once in six months and Pet CT or MRI screening after a year of which MRI Abdomen was done which revealed no change in cysts and no new malignant lesions.

DISCUSSION

In this particular case the patient presented with multiple cysts in pancreas, kidney and adrenal gland. The main concern was pain abdomen. By the time she reached us, she already delivered a baby, had undergone partial nephrectomy. But she had undergone a diagnostic dilemma before reaching us. With careful past history and well directed investigations, we were able to pin point the diagnosis of VHL. We were able to manage the case with conservative management. Neupane et al reported a case of VHL in 50-year-old male patient.[5] He mainly had lesions in the central nervous system, retina and cortex of kidney. They managed the case surgically. The patient responded well to the treatment and was free of signs and symptoms at three months of surgery. Khanduri et al noted the VHL in male siblings.[6] Both the kidneys were involved with multiple cysts in pancreas. The 32-year-old male patient of this case first presented with pain abdomen. Signs and symptoms were suggestive of mass in the abdomen and later confirmed by investigations. Here, the authors did not describe further follow up of this case.

Fan et al described a case of VHL in a woman. She was detected to have mutation c.353T > C in exon 2 of the short arm of chromosome 3.[7] Her two family members were also diagnosed with same mutations. But the clinical presentation was different in every case. The index case had hemangioblastoma of central nervous system, RCC. She was surgically treated and had a better prognosis. Hemangioblastomas of central nervous system (CNS) may be seen in 60-80% of VHL cases. Mutations of exon and intron may cause CNS hemangioblastomas. Symptoms depend upon location of tumor. Surgical resection is the treatment of choice but may be a challenge. There can be recurrence. Surgery combined with radiotherapy can have a five-year survival rate of 80-90%.[8]

About 70% of the cases present with pancreatic lesions as seen in this case report. Cysts in the pancreas are more common. In general population, pancreatic cysts are not common. Hence, anyone having pancreatic cyst should be suspected to have VHL and should be evaluated in that direction. Only suspected malignancy cases should be

managed with surgery. Others can be managed conservatively as in the present case.[9] Renal lesions can be seen in 60% of cases with VHL. Renal cell carcinoma (RCC) and renal cysts presentation is common. RCC is major cause of death. Renal mass more than 3cm should be considered for surgery.[10]

CONCLUSION

We conclude that we came across a 27-year-old female patient with pain abdomen as main presenting complaint. Specific diagnosis of this syndrome was not possible in her previous consultations at some other clinics. But, when she presented to use, we thoroughly investigated her in right direction and diagnosed with VHL. She responded to the conservative treatment and reported to be symptom free at one year follow up.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Chappell JC, Payne LB, Rathmell WK. Hypoxia, angiogenesis, and metabolism in the hereditary kidney cancers. J Clin Invest. 2019;129(2):442-51.
2. Tsang SH, Sharma T. Von Hippel-Lindau Disease. Adv Exp Med Biol. 2018;1085:201-3.
3. van Leeuwaarde RS, Ahmad S, van Nesselrooij B, Zandee W, Giles RH. Von Hippel-Lindau Syndrome. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, editors. GeneReviews. University of Washington, Seattle; Seattle (WA). 2000.
4. Koch CA, Petersenn S. Black swans-neuroendocrine tumors of rare locations. Rev Endocr Metab Disord. 2018;19(2):111-21.
5. Neupane D, Dahal A, Lageju N, Jaiswal LS, Kandel A, Manandhar S. Von Hippel-Lindau disease: A case report. Int J Surg Case Rep. 2022;97:107417.
6. Khanduri S, Khan N, Malik S, Katara S, Fatima M. Von Hippel-Lindau disease: a rare radiological case report of a symptomatic patient and his asymptomatic genetic counterpart. Cureus. 2021;13(1):12925.
7. Fan X, Wang S, Chen T, Hu W, Yang H. Von-Hipple Lindau syndrome with family history: a case report and seventeen years follow-up study. Front Oncol. 2024;14:1360942.
8. Larcher A, Belladelli F, Fallara G, Rowe I, Capitanio U, Marandino L, et al. Multidisciplinary management of patients diagnosed with von Hippel-Lindau disease: A practical review of the literature for clinicians. Asian J Urol. 2022;9:430–42.
9. Blansfield JA, Choyke L, Morita SY, Choyke PL, Pingpank JF, Alexander HR, et al. Clinical, genetic and radiographic analysis of 108 patients with von Hippel-Lindau disease (VHL) manifested by pancreatic neuroendocrine neoplasms (PNETs). Surgery. 2007;142:814–8.
10. Sacco E, Pinto F, Sasso F, Racioppi M, Gulino G, Volpe A, et al. Paraneoplastic syndromes in patients with urological malignancies. Urol Int. 2009;83:1–11.

Cite this article as: Avva VS, Reddy DN. Von Hippel-Lindau syndrome: a rare case report from a tertiary care hospital. Int J Adv Med 2025;12:516-8.