Case Report

Radiological Spectrum of von Hippel-Lindau disease – A Case Report

Sharma S[1], Sharma S[2], Ramchandran P[3], Sharma S[4], Sharma BB[5]

From 1Department of Pediatrics, and 2Department of Anaesthesia, SGT Medical College, Gurgaon, India, 3Department of Anesthesia, Heartlands Hospital, Brimingham, UK, 4 Department of ENT, Head & Neck Surgery, and 5Department of Radio diagnosis, Post Graduate Institute of Medical Education and Research & Dr. Ram Manohar Lohia hospital, New Delhi, India.

Correspondence to: Dr Shashi Sharma, B-32, Nivedita Kunj, Sector 10, R K Puram, New Delhi – 110022. E mail: shashi.s986@gmail.com. Tel No +91 11 26712700

Received: 18 December 2014 Initial Review: 07 February 2015 Accepted: 23 February 2015 Published online: 28 February 2015

ABSTRACT

Von Hippel-Lindau (VHL) disease is an autosomal dominant genetic disorder resulting from inactivation of tumor suppression genes located at the chromosome 3p 25.5. VHL comprises of many benign and malignant tumors along affecting various systems of the body with variable manifestations. A wide variation in radiological spectrum is seen as per the involvement of particular system or organ. The imaging modalities like Ultrasonogrphy (USG), Computerized Tomography (CT) and Magnetic Resonance Imaging (MRI) play an important role in diagnosing and treating the affected individuals. These also play equally important role in the screening and follow up of such cases. We present a case of an 18 year old female, who was diagnosed with VHL at the age of 10 years, where imaging modalities helped her management and follow up.

Keywords: *VHL, autosomal dominant, genetic disorder, USG, CT, MRI*

Von Hippel-Lindau disease (VHL) was named after a German Ophthalmologist Eugen von Hippel and Swedish pathologist Arvid Vilhelm in 1964. This is an autosomal dominant genetic disorder resulting from inactivation of tumor suppression genes located at the chromosome 3p 25.5.This is a multisystem disorder and has got variable clinical manifestations such as hemangioblastomas of the retina and central nervous system, renal tumors and cysts, pancreatic tumors and cysts, pheochromocytomas, endolymphatic sac tumors and epididymal cystadenomas. The mutation results in 20% of the cases. The prevalence of the disease is 1 in 31,000 to 1 in 53,000.

CASE REPORT

An 18 year old female reported with the complaints of headache, anxiety, abdominal pain and abdominal distension. She was diagnosed as a case of von Hippel-Lindau disease at the age of ten year when she presented with similar complaints. Diagnosis was made on the basis of biochemical parameters including VMA levels, USG, CECT and MRI findings. There was no family history suggestive of the disease.

At that time, abdominal USG suggested possibility of VHL by showing huge pancreas studded with multiple cysts which was later supported by CECT abdomen. MRI head revealed multifocal hemangioblastomas in cerebellum which was pressing upon the fourth ventricle resulting in features of raised intracranial pressure and headache. Surgical management led to relief in the ongoing obstructive hydrocephalic pathology as well as the confirmation of diagnosis by histo-pathological examination.

At present the biochemical parameters are within the normal range including equivocal VMA level. Other investigations have shown as Hb 10g/dL and BP was 150/100 mm of Hg. Now the patient is on constant follow

up to see the behavior of the disease and to proceed for the management accordingly. The patient was reviewed with radiological investigations by USG, CT and MRI. USG abdomen has shown the enlarged pancreas studded with multiple simple cysts. Both the kidneys show multiple small masses of mixed echogenecity.

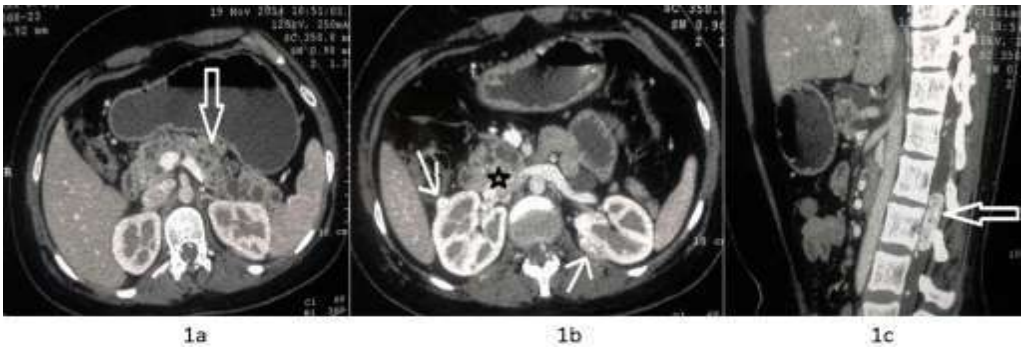

Figure 1 - CECT Abdomen, (a) Axial section shows the enlarged pancreas studded with multiple cysts (wide white arrow). (b) Axial section shows tumors in both the kidneys (white arrows). The head of pancreas is also seen full of cysts (black star). (c) Sagittal reformatted image shows the enhancing tumor in the spinal cord (white wide arrow).

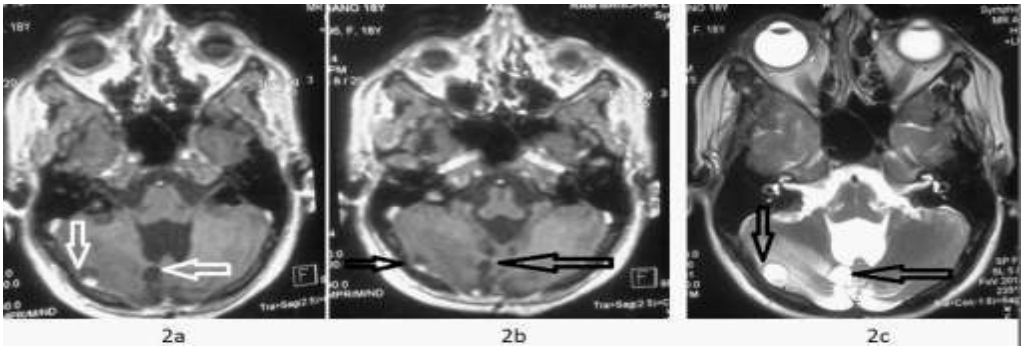

Figure 2 - MRI Brain Axial sections. (a) Multiple enhancing hemangioblastomas in the cerebellum (white arrows). (b) Another section shows similar type of lesions (small black arrow).One of the small tumor has been surgically removed (large black arrow on left side). (c) T2W image show multiple hyperintense cystic components of the tumor (black wide arrows).

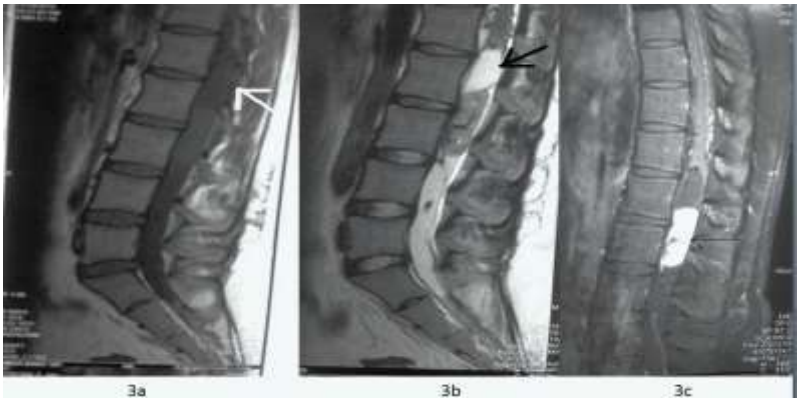

Figure 3 - MR Dorsolumbar spine (a) T1W Sagittal section shows iso to hypointense lesion in the cord (white arrow). (b) T2W Sagittal section shows hyperintensity in the cystic component (black arrow) and hypointensity in the solid component of the lesion.(3c) Contrast enhanced T1W Sagittal section shows intense contrast enhancement in the solid component of the hemangioblastoma within the cord (black arrow) leaving cystic component unchanged.

CECT head and abdomen has shown multiple hemagioblastomas in cerebellum, kidneys and spinal cord (Figure 1 a,b and c). The pancreatic parenchyma was largely replaced by multiple cysts of fluid density. Brain MRI also shows multiple cerebellar hemagioblastomas (2 a,b and c) and multiple bilateral tumors in kidneys .MRI of the spine has shown the solid cystic tumor in the dorso lumbar region (Figure 3a and b).Contrast MRI has shown intense enhancement of the solid component (3 c). There was also tethering and compression of the cord.

DISCUSSION

VHL is a multisystem disorder which requires correct early diagnosis and management. Many patients develop end stage complications because of the underlying renal and neurological involvement [1,2]. Following manifestations are in the order of the involvement [3]: Pancreatic cysts (50-91%), Cerebellar hemangioblastoma (44-72%), Renal cysts (59-63%), Retinal hemangioblastoma (45-59%), Renal cell carcinoma (24-45%), Spinal cord hemangioblastoma (13-59%), Pheochromocytoma (0-60%), Neuroendocrine tumor of the pancreas (5-17%), Serous cystadenoma of the pancreas (12%), Medullary hemangioblastoma (5%), Papillary cystadenoma of the epididymis (10-60%).

These cases can be classified into two categories: Type I has got very low risk of pheochromocytoma but Type II has got additional risk of renal cell carcinoma (RCC).Family history is very important otherwise minimal two or more CNS or retinal hemagioblastomas with other visceral tumors confirm the diagnosis. The patient presents with symptoms almost like that of tumor.

Leung et al [4] has advised the screening protocol as follow: 1) Annual blood pressure and neurological examination. 2) Annual ophthalmoscopy from 5 years of age. 3) Annual 24 –hour estimation of vanillylmandelic acid (VMA) from 10 years of age. 4) Annual ultrasound examination of the abdomen since 10 years of age. 5) Baseline Magnetic Resonance imaging examination of brain and spine at age of 20 years. 6) MRI examination if Audiogram is positive.

Chokye PL et al (1995) have shown that CNS hemangioblastoma is the most common manifestation of this entity [6]. The average age in these patients is 50 years and the cause of mortality is mainly because of renal cell carcinomas and CNS hemagioblastomas.Malek RS et al (1987) have revealed that CNS hemangioblastoma usually precedes the renal disease. The radiological imaging plays a vital role in diagnosing many entities of the spectrum. These also play a key role in screening and follow up of the diagnosed cases. The management can be tailored by early detection of the various manifestations. The imaging modalities like USG, CECT and contrast enhanced MRI guide the management as per the severity of the disease according to the protocol and requisition. USG reveals quickly about the character of the intra abdominal lesion in their consistency and location. The patient is subjected to the next modality as per the requirement.

The majority of the pancreatic neuroendocrine tumors are small and located in the head region which enhance homogenously if solid in consistency. These are prone to metastasize if the size is more than 3.0 cm .CECT and contrast MRI depict with heterogeneous enhancement pattern [5]. MRI has got advantage being superior in tissue characterization and of non radiation in nature. The hidden lesions like retinal hemangioblastomas can easily be picked up by this modality [6]. There has to be multidisciplinary approach for the management depending upon the presenting complaints. The surgical excision is the best solution if the patient is symptomatic and follow up should be adhered like it was done in our case..

CONCLUSION

VHL is multisystemic disorder which requires correct diagnosis, monitoring and management. This is best achieved by the right follow up as per the protocol. The mainstay of this follow up is by radiological imaging modalities like USG, CT and MRI. In our present case the patient was operated upon for the posterior fossa hemangioblastoma as this was leading to dominant symptomatology. Later on the progression of various tumors were monitored by the various imaging modalities.

REFERENCES:

1. Taouli B, Ghouadni M, Correas JM Hammel P, Couvelard A, Richard S, et al. Spectrum of abdominal imaging findings in von Hippel-Lindau disease. AJR Am J Roentgenol. 2003;181(4):1049-54.
2. Courcoutsakis NA, Prassopoulos PK, Patronas NJ. Aggressive leptomeningeal hemangioblastomatosis of the central nervous system in a patient with von Hippel-Lindau disease. AJNR Am J Neuroradiol. 2009;30(4):758-60.

3. Karsdorp N, Elderson A, Wittebol-Post D, Hene RJ, Vos J, Feldberg MA, et al. Von Hippel Lindau disease: new strategies in early detection and treatment. Am J Med. 1994;97(2):158-68.
4. Leung RS, Biswas SV, Duncan M, et al. Imaging features of von Hippel-Lindau disease. Radiographica. 2008;28(1):65-79.
5. Marcos HB, Libutti SK, Alexander HR, Lubensky IA, Bartlett DL, Walther MM, et al. Neuroendocrine tumors of the pancreas in von Hippel-Lindau disease: Spectrum of appearances at CT and MRI imaging with histopathologic comparison. Radiology. 2002;225:751-8.
6. Choyke PL, Glenn GM, Walther MM, Patronas NJ, Linehan WM, Zbar B. Von Hipple-Lindau disease: Genetic, clinical, and imaging feature. Radiology. 1995;194:629-42.

How to cite this article: Sharma S, Sharma S, Ramchandran P, Sharma S, Sharma BB. Radiological Spectrum of von Hippel-Lindau disease – A Case Report. Indian J Case Reports. 2015;1(1):14-17.

Conflict of interest: None stated, Funding: Nil