

# Von Hippel Lindau Disease [VHL]: Magnetic Resonance Imaging Spectrum in a Single Patient

*Sandeep G. Jakhere, Bhakti Yeragi, Darshan G. Jain*

Department of Radiology, B Y L Nair Charitable Hospital and T N Medical College, Dr A R Nair Road, Mumbai Central, Mumbai, Maharashtra, India – 400008. Correspondence mail: drsandeepjakhere@gmail.com.

## ABSTRAK

*Artikel ini merupakan suatu laporan kasus yang menjelaskan spektrum pencitraan resonansi magnetik (MRI) yang luas pada seorang pasien dengan keterlibatan organ multipel. Penyakit Von Hippel Lindau merupakan kelainan autosomal dominan yang diturunkan dan ditandai dengan lesi jinak dan ganas yang melibatkan berbagai organ.*

*Pencitraan memainkan peranan penting dalam penegakan diagnosis dan pengamatan (surveillance) VHL karena dapat membedakan lesi jinak dan lesi ganas. Nilai median usia harapan hidup penderita VHL adalah 49 tahun dan penyebab kematian yang paling sering adalah karsinoma sel ginjal dan komplikasi neurologik akibat hemangioblastoma serebelum.*

**Kata kunci:** *penyakit Von Hippel Lindau, hemangioblastoma, karsinoma sel ginjal, kista ginjal.*

## ABSTRACT

*In this case, we describe the wide magnetic resonance imaging (MRI) spectrum in a single patient who had multi organ involvement. Von Hippel Lindau Disease is a rare inherited autosomal dominant disorder characterized by development of benign and malignant lesions involving multiple organs.*

*Imaging plays a vital role in the diagnosis and surveillance of VHL as it can differentiate benign from malignant lesions. The median life expectancy of VHL is 49 years and the commonest cause of death in patients with VHL is renal cell carcinoma and neurologic complication from cerebellar hemangioblastomas*

**Key words:** *Von Hippel Lindau disease, hemangioblastoma, renal cell carcinoma, renal cyst.*

## INTRODUCTION

Von Hippel Lindau Disease is a rare inherited autosomal dominant disorder characterized by development of benign and malignant lesions involving multiple organs. Imaging plays a vital role in its evaluation and for disease surveillance. We describe the wide magnetic resonance imaging spectrum in a single patient who had multi organ involvement.

## CASE ILLUSTRATION

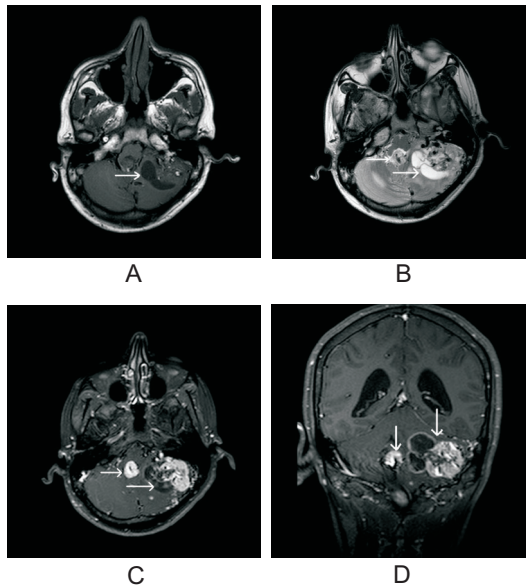
A 28 year old male presented with complaints of persistent occipital headache and vomiting for the last 20 days. He also complained of swaying

to left side since last 8 months. His father expired few years back due to renal cell carcinoma. On examination there was papilledema in the left eye and cerebellar signs were positive i.e. on tandem walking there was swaying to the left side. A clinical diagnosis of left cerebellar space occupying lesion with raised intracranial tension was considered and a MRI was requested.

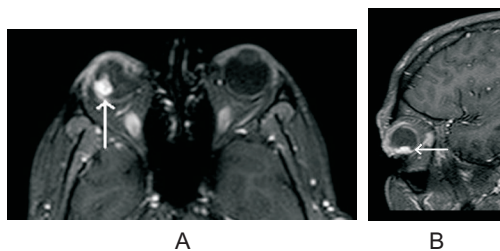
MRI brain revealed a fairly well defined intra axial lesion within in the left cerebellar hemisphere which appeared iso to hypointense on T1, heterogeneously hyperintense on T2 and showed an irregular moderate enhancement on the post contrast images. The eccentric cystic portion appeared hypointense on T1, hyperintense on T2

and showed a peripheral enhancement on the post contrast images. Multiple flow voids were seen within the lesion confirming the hyper vascular nature of the lesion. There was mild perilesional edema Few solid enhancing lesions were also noted in posterior portion of left cerebellar hemisphere, right cerebellar hemisphere, vermis, base of 4th ventricle. **(Figure 1)** A well defined solid lesion with intense enhancement was seen in posterior chamber of right globe. **(Figure 2)**

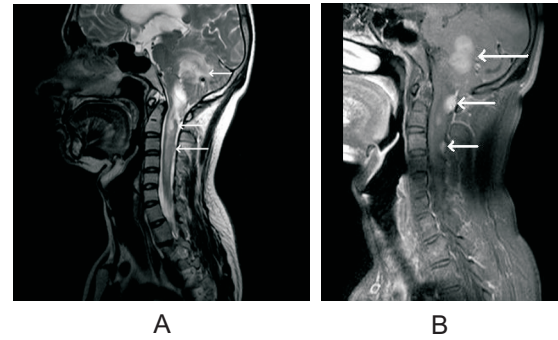
MRI spine revealed diffuse signal abnormality in the upper cervical cord with moderate cord swelling on T2 weighted images **(Figure 3)**. On post gadolinium images three solid enhancing lesions were seen in upper cervical cord. **(Figure 3(b))**



**Figure 1.** Intra axial lesion within the left cerebellar hemisphere which appears. (a) iso to hypointense on T1W, (b) heterogeneously hyperintense on T2W, (c, d) shows irregular moderate enhancement on the post contrast images. The eccentric cystic portion appears (a) hypointense on T1, (b) hyperintense on T2 and shows peripheral enhancement on the post contrast images. Multiple flow voids are seen within the lesion. Solid enhancing lesions also noted in (c) and (d) vermis, base of 4th ventricle



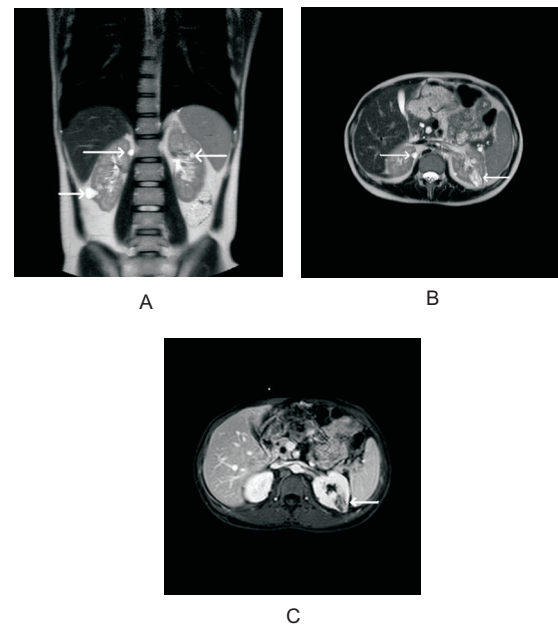
**Figure 2.** A well defined solid lesion with intense enhancement seen in posterior chamber of right globe. Solid enhancing lesions also noted in posteroinferior portion of right cerebellar hemisphere



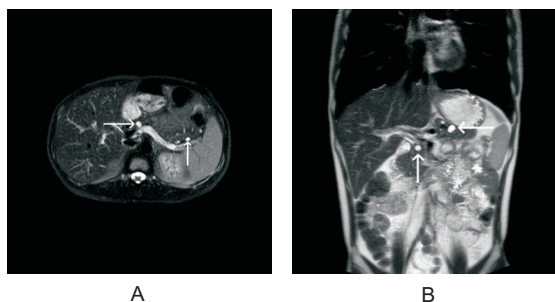
**Figure 3.** (a) Diffuse signal abnormality in the upper cervical cord with moderate cord swelling on T2W images. On post gadolinium images (b) three solid enhancing lesions seen in upper cervical cord.

In view of the history and the imaging findings suggestive of multiple haemangioblastomas, a possibility of von Hippel Lindau disease was considered and a MRI screening of the abdomen was performed.

MRI abdomen revealed multiple simple cysts in both kidneys **(Figure 4 (a))**. A larger cyst showed a soft tissue component which showed heterogeneous enhancement on the post contrast images raising a possibility of a renal cell carcinoma. **(Figure 4(b), 4(c))**. Multiple simple cysts were also seen within the head and the tail of the pancreas. **(Figure 5(a), 5(b))**.



**Figure 4.** (a) T2W axial and coronal images show multiple simple cysts in both kidneys. (c) Larger cyst in left kidney shows a soft tissue component with heterogeneous enhancement on the post contrast images.



**Figure 5.** Multiple simple cysts seen within the head and the tail of the pancreas.

## DISCUSSION

Von Hippel Lindau disease is a rare multisystem disorder characterized by development of multiple benign and malignant tumors in the brain, spinal cord, pancreas, kidney, adrenals and the epididymis. It has an autosomal dominant inheritance with a high penetrance and variable expression. There is a 50% chance of a inheriting the VHL gene from a carrier.<sup>1</sup> The prevalence of VHL ranges from one in 39000 to 53000 and is associated with inactivation of a tumor suppression gene on short arm of chromosome 3.<sup>2</sup>

The clinical diagnostic criteria were proposed by Melmon and Rosen in 1964<sup>3</sup> and include 1). More than one CNS hemangioblastoma; 2). CNS hemangioblastoma and visceral manifestation of VHL disease; 3). Any manifestation and a known family history of VHL disease.

CNS hemangioblastomas are seen in 60-80% of patients with VHL<sup>4</sup> and they usually have a predominantly cystic appearance with a enhancing mural nodule on MRI. Because of the hyper vascular nature of the tumor, multiple flow voids are usually seen on T1 and T2 weighted images and is characteristic of the lesion. The common sites are the cerebellum, spinal cord and the medulla.

Renal cysts are usually seen in 59-63% of patients with VHL<sup>1</sup> and are usually simple cysts with the typical appearance of being hypointense on T1, hyperintense on T2 and negligible post contrast enhancement. However, presence of thick septations, enhancing soft tissue component or chunky calcification may raise a possibility of a RCC which occurs in 24-45% patients with VHL.<sup>5</sup>

Pancreatic lesions are seen in 35-70%<sup>4</sup> of patients and may manifest as simple cysts or pancreatic tumors, the commonest being pancreatic neuro endocrine tumors and pancreatic cystadenoma. Imaging of pancreatic neuro endocrine tumors may require specific imaging in the pancreatic phase of enhancement as these lesions are known to be conspicuous in nature.

Phaeochromocytomas arise in 10-20% of patients with VHL<sup>4</sup> with a mean age of presentation of 30 years. Diagnosis is based on a combination of imaging and laboratory investigation (plasma free metanephrine).

In addition to the above lesions, epididymal cystadenomas and broad ligament cyst adenomas may also be seen.

## CONCLUSION

Imaging plays a vital role in the diagnosis and surveillance of VHL as it can differentiate benign from malignant lesions. The median life expectancy of VHL is 49 years and the commonest cause of death in patients with VHL is renal cell carcinoma and neurologic complication from cerebellar hemangioblastomas.<sup>1</sup>

## REFERENCES

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