ORIGINAL RESEARCH PAPER

Radiodiagnosis

KEY WORDS: Von Hippel-Lindau (VHL), Hemangioblastomas, Central Hemangioblastomas (CHbs), pancreatic cyst, pancreatic neuroendocrine tumor, b/l renal cell carcinoma

RARE CASE PRESENTAETION: VON HIPPLE LINDAU SYNDROME

Dr. Bhaumik Jayeshbhai patel 3rd year resident at department of radiodiagnosis, Gujrat cancer research institute, Asarwa, Ahmedabad- 380016 Gujarat state, India

Dr. Akilesh Bharatbhai Patel* senior resident at department of radiodiagnosis, civil hospital, B J medical college, Asarwa, Ahmedabad- 380016 Gujarat state, India*Corresponding Author

Dr. Viraj Bababhai Shah 3rd year resident at department of radiodiagnosis, Gujrat cancer research institute, Asarwa, Ahmedabad- 380016 Gujarat state, India

Dr. Himanshu soni MD DMRE, Professor department of radiodiagnosis, Gujrat cancer research institute, Asarwa, Ahmedabad- 380016 Gujarat state, India

ABSTRACT

Introduction: Von Hippel-Lindau syndrome (VHL) is autosomal dominant hereditary syndrome due to germline mutations of the tumor suppressor gene VHL, located on short arm of chromosome 3. 80% have a positive family history and about 20% of cases are de novo. Various multiple benign and malignant tumors occur affecting multiple organ system, central nervous system, kidneys, adrenals, pancreas, and reproductive system. Common manifestations include including retinal hemangioblastomas (HBs), central nervous system (CNS) HBs, endolymphatic sac tumors, pancreatic neuroendocrine tumors, pancreatic cystadenomas, pancreatic cysts, clear cell renal cell carcinomas, renal cysts, pheochromocytomas, paragangliomas, and epididymal and broad ligament cystadenomas. Confirmation of diagnosis is done by genetic testing in clinically and radiologically suspicious patient. **Case Report:** A male patient of age 30 years presented with cerebellar symptoms 2 year ago and MRI is done MRI reveal cerebellar and spinal hemangioblastoma. Patient is operated for cerebellar hemagioblastoma and histologically confirmed. On follow up study b/l renal cell carcinoma, pancreatic cyst and neuroendocrine tumor is found. **Conclusion:** VHL is a disease with no cure. Regular follow-up with necessary to follow the previous lesions and detect any newly developed VHL-associate tumors.

INTRODUCTION:

Von Hippel–Lindau syndrome (VHL) is hereditary autosomal dominant syndrome, due to mutations in the VHL tumor suppressor gene, located on the short arm of chromosome 3 when both copies of gene is inactivated lead to unregulated cell growth. The gene has high penetrance but variable expression[3].

Prevalence: 1:36000[1]

Around 80% cases are hereditary and 20 % cases arises denovo. The mean age of onset of 26 years and 97% of people with a VHL gene mutation have symptoms by the age of 65. Patients develop multiple benign and malignant tumors involving various multiple organ systems[2]

Manifestation ofVHL

CNS

Retinal, cerebellar or spinal Hemangioblastoma Choroid plexus papilloma

Retinal hemangioblastoma also known as retinal angioma. On nonenhanced T1-weighted MR imaging, lesions demonstrate higher signal intensity than normal vitreous. Significant contrast enhancement is seen only in the most severely affected patients of these patient has profound loss of vision.

Urogenital system

Renal lesions:

Renal cyst: Often bilateral and multiple may be simple or complex type Renal cell carcinoma: Usually clear cell type Renal angiomyolipoma

Genital lesion

Epididymal cyst
Papillary cystadenoma of epididymis
Broad ligament cystadenoma

Pancreas:

Pancreatic cyst
Neuroendocrine tumor of pancease
Serous cystadenoma of pancrease
Pancreatic adenocarcinoma

Liver: cyst
Adrenal: Pheochromocytoma

Endolymphatic sac tumor
Extraadrenal pheochromocytoma

Diagnostic criteria forVHL

1. More than one CNS hemangioblstoma
2. One CNS hemangioblastoma and visceral manifestation of VHL
3. Any manifestation and known family history

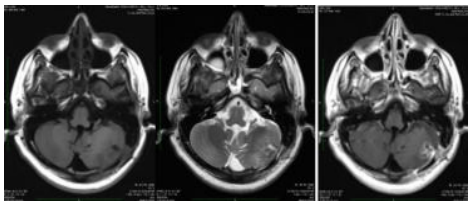
CASE PRESENTATION

A 30-year-old male patient presented with symptoms of headache and tandem walking, dysdiadokokinesia. Magnetic resonance imaging of brain and cervicodorsal spine was done which revealed a mass in the left cerebellum and cervical and upper dorsal spine which is extending from cervico-medullary junction to D3 vertebral level.

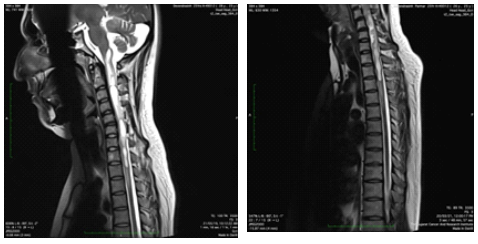
IMAGING FINDINGS

On MRI lesion is predominantly hypointense in nature with solid component appear isointense on T1 images on T2W images lesion appear hyperintense with vascular flow void in peripheral part of lesion. On post contrast imaging solid component of lesion shows enhancement. (Figure 1,2,3)

Figures 1,2,3



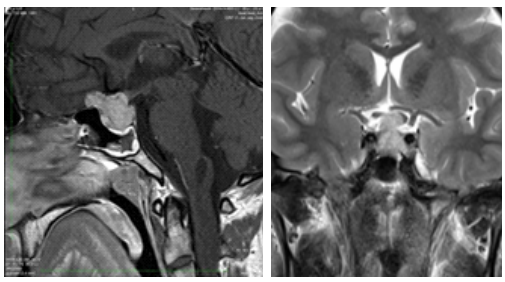
In spinal cord multi-cystic lesion is noted which is extending from corticomedullary junction to D3 vertebral level which appear hypointense on T1W images and hyperintense on T2W images solid component of lesion at C2 and C6 vertebral level shows intense post contrast enhancement. There is associated syrinx formation is noted. There is mild hyperintensity is noted in spinal cord at D4 vertebral level on STIR imaging. S/o oedema. (Figure 4,5)



Figures 4, 5

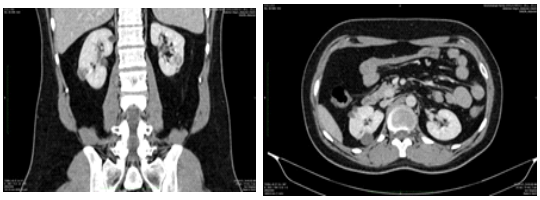
Patient is operated for mass in left posterior fossa and histopathologically proven as hemangioblastoma.

In MRI brain there is altered signal intensity lesion is seen which appear hypointense on T1W imaging and appear hyperintense on T2 imaging, lesion extend into suprasellar cistern. Optic chiasma not seen separately from lesion. Lesion extend into cavernous sinus region on both sides. Lesion abuts left intracavernous ICA with arch of contact < 180 degree and encases right intracavernous ICA with arch of contact >180 degree. Lesion shows internal small cystic area. Lesion causes erosion of floor of sella and extend to left sphenoid sinus. Lesion is snowman/ figure of 8 in appearance. These finding are suggestive of pituitary macroadenoma. (Figure 6,7)



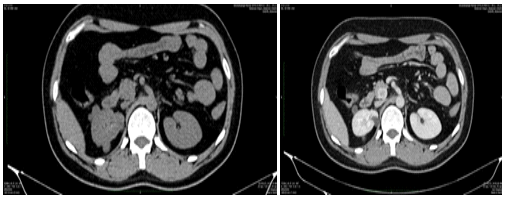
Figures 6, 7

On follow up study on CT there is multiple cyst is noted in both kidney and on post contrast study heterogeneously enhancing mass is noted in kidney on both side which causes contour deformity. (Figure 8,9)



Figures 8, 9

On USG pancreases hypoattenuating lesion is noted in uncinate process of pancreas which on post contrast CT shows intense enhancement. (Figure 10,11)



Figures 10, 11

There is cystic lesion is noted in head of pancreas which is anechoic on ultrasound and shows fluid attenuation.

Conclusion:

VHL is disease with no cure till date. Most common death result from renal cell carcinoma and neurological symptoms resulting from cerebellar and spinal hemangioblastoma.

Regular follow-up with imaging is necessary to follow the previous lesions and detect any newly developed VHL-associate tumors. Pharmacological treatment is not available early detection and surgical removal of tumor improve patient life expectancy.

Early detection and treatment of tumor significantly improve patient diagnosis.

VHL can't be prevented once patient have.
Genetic counselling should be provided to patient having VHL.
VHL Alliance Surveillance Recommendations

At birth	Neurological examination, ophthalmic examination, new born hearing test
1-4 year	yearly ophthalmic examination and clinical evaluation for neurologic disturbances, blood pressure, and hearing
5-15 year	All of the surveillance recommendations at birth and at ages 1–4. Annually biochemical tests, including plasma metanephrine, abdominal US from 8 years of age or earlier if indicated; MR imaging of the abdomen or functional imaging with MIBG scintigraphy to be performed only if biochemical abnormalities are found
>16 year	Audiology assessment every 2–3 years annually if any auditory-vestibular symptoms present MR imaging with contrast enhancement of the internal auditory canal every 2–3 years Annual comprehensive ophthalmic examination Annual clinical evaluation for neurologic disturbances.

REFERANCE

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