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Original Research Paper

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RARE CASE OF VON HIPPEL-LINDAU SYNDROME

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ABSTRACT Von Hippel–Lindau (VHL) disease is a rare, autosomal dominantly inherited multisystem disorder characterized by development of a variety of benign and malignant tumors. We report a rare case of Von Hippel Lindau Syndrome and discuss about the Clinical Presentation and Neurosurgical Management of the patient with Von Hippel Lindau Syndrome. Due to a combination of robust radiologic and clinical screening and advanced surgical techniques, the morbidity and mortality of patients with VHL disease has been reduced significantly. A Multidisciplinary team approach is important in screening for VHL disease.

KEYWORDS : Von Hippel-Lindau Syndrome, Cervical Hemangioblastoma , Pancreatic Cyst, Retinal Hemangioma

INTRODUCTION

Von Hippel–Lindau (VHL) disease is a rare, autosomal dominantly inherited multisystem disorder characterized by development of a variety of benign and malignant tumors. [1, 2, 3]

The spectrum of clinical manifestations of the disease is broad. These include retinal and central nervous system (CNS) hemangioblastomas, endolymphatic sac tumors, renal cysts and tumors, pancreatic cysts and tumors, pheochromocytomas, and epididymal cystadenomas.

CASE REPORT :

41 year old Gentleman presented with Weakness of right hand of 2 months duration and Weakness of right lower limb of 2 months duration with H/o Neck pain present for 1 ½ months duration; Patient had Drooping of right upper eyelid for 1 ½ months duration. History of Multiple Small Swellings all over the body. Family History present for multiple small swellings all over the body. Multiple swellings in forearm and trunk clinically suggestive of Lipoma. Bilateral hydrocele present. The patient had asymmetrical Progressive Quadriparesis (Right more than left) with sensory disturbances and drooping of Right Upper eyelid. On Examination: Right eye had partial ptosis. Ciliospinal reflex absent on right side. Pupils Right 2.5 mm RTL Left pupil 3 mm RTL. Fundus Examination revealed Super temporal quadrant elevated red lesion in left eye suggestive of hemangioma. Spino motor examination revealed Pyramidal Type of weakness. Weakness exaggerated in lower limbs. Abdominal Reflexes absent in right quadrant. Sensation C 2 and below C 2 diminished.

Diagnostic Investigations :

MRI of Cervical Spine with whole spine screening showed cervical Hemangioblastoma of size 1.4 (CC) x 0.7 (AP) x 1.4 (T) cm well defined homogenously enhancing intramedullary lesion noted at C 6 level with associated cord expansion and adjacent pial enhancement and peritumoral edema. MRI Brain had no significant abnormalities.

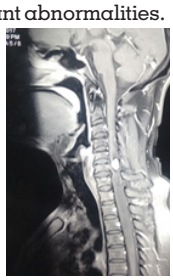

Figure 1: Preoperative MRI Sagittal with Contrast

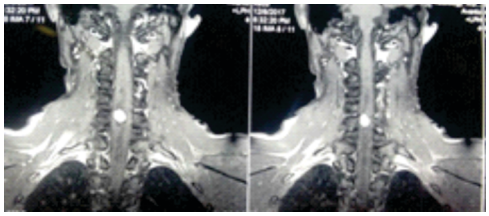

Figure 2: Preoperative MRI Coronal with Contrast

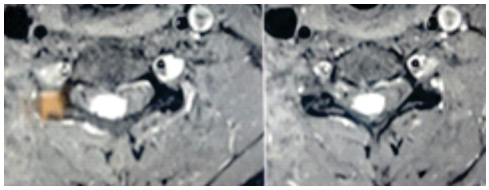

Figure 3: Preoperative MRI Axial with Contrast

Non enhancing non tumoral cysts noted proximal to the lesion extending from C 3 to C 6 level and also distal to the lesion extending from C 7 to D 5 level.

CT Abdomen imaging was done.

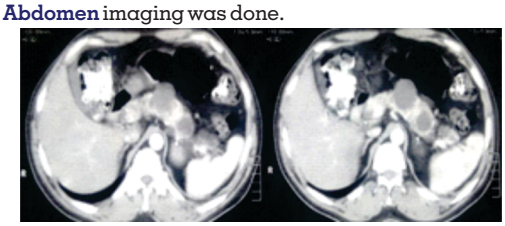

Figure 4: CT Abdomen with Pancreatic Cysts.

Imaging Impression: F/S/O Von Hippel-Lindau Syndrome

Left Eye FFA revealed Hemangioma in Left superotemporal Quadrant.

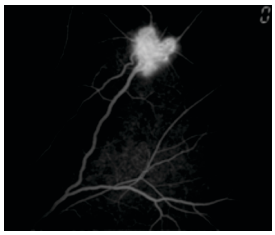

Figure 5: Left eye FFA Findings

Treatment : Patient underwent cervical laminectomy and total Excision of C 6 level intramedullary SOL .

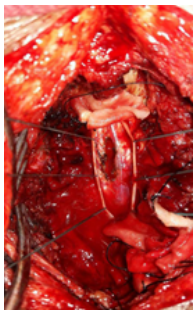

Figure 6: Intraoperative Picture

HPE suggestive of Hemangioblastoma Post operatively weakness and sensory symptoms improved.

Post operative MRI Study of Cervical Spine with WSS – Plain & Contrast was done which showed no residual lesion.

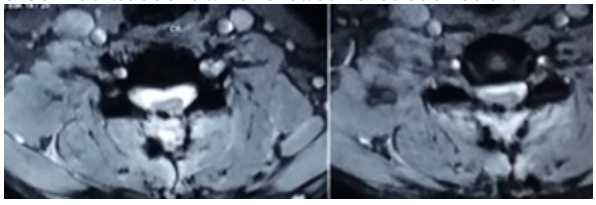

Figure 7: Post-operative – MRI T 2 Axial images with no residual lesion

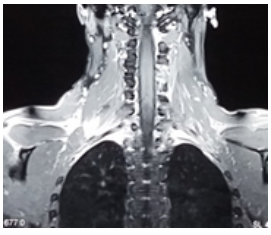

Figure 8: Post-operative – MRI Contrast Coronal Image with no residual lesion

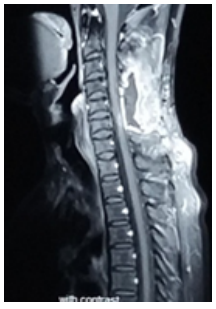

Figure 9 : Post-operative – MRI Contrast Sagittal Image with no residual lesion

After discharge patient referred for Ophthalmic Institute for Laser Photocoagulation of Retinal Hemangioma. At 2 and 6 months follow up patient had improved symptomatically.

DISCUSSION

Von Hippel–Lindau (VHL) disease is a rare, autosomal dominantly inherited multisystem disorder characterized by development of a variety of benign and malignant tumors. [1, 2, 3]

The inheritance is autosomal dominant; that is, there is a 50% chance of inheriting the VHL gene from a carrier. The gene has high penetrance but variable expression, resulting in a wide variety of manifestations of the disease in affected individuals.

The VHL gene is a tumor suppressor gene; thus, when both copies of the gene are inactivated by mutation or loss, cell growth is unregulated and tumors in multiple organs result. The location of the tumor suppression gene was isolated to chromosome 3p25.5 [2, 3]

The spectrum of clinical manifestations of the disease is broad. About 40 different lesions in 14 different organs have been described . These include retinal and central nervous system (CNS) hemangioblastomas, endolymphatic sac tumors, renal cysts and tumors, pancreatic cysts and tumors, pheochromocytomas, and epididymal cystadenomas.

Spinal Hemangioblastomas occur in 13%–50% of VHL Cases. The onset is reported at a median age of 33 years and the literature review gives a range of 11–66 years. Spinal Hemangioblastomas are more common in the thoracic and cervical cord. They manifest with signs & symptoms of radiculopathy or myelopathy like hyperesthesia, weakness, gait ataxia, hyperreflexia, pain, incontinence, and rarely quadriplegia [4].

Retinal Hemanigioblastomas, also called retinal angiomas or capillary hemangiomas, seen in 45%–60% of VHL cases [5,6].

Pancreatic cysts occur frequently in VHL. Literature reveals that pancreatic cysts occur in a range of 7 % to 72 % of VHL cases. Pancreatic cysts are typically multiple and mostly asymptomatic and may be the only manifestation at the time of initial diagnosis of VHL [7].

The most common causes of death in VHL disease patients are renal cell carcinoma and neurologic complications from CNS hemangioblastomas.

CONCLUSION

Due to a combination of robust radiologic and clinical screening and advanced surgical techniques, the morbidity and mortality of patients with VHL disease has been reduced significantly. Screening is important especially with those having family history because the lesions in VHL disease are treatable [8, 9] . Thus, early detection allows use of more appropriate therapy and may enhance the patient's length and quality of life. A Multidisciplinary team approach is important in screening for VHL disease.

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