CASE REPORT

CEREBELLAR HEMANGIOBLASTOMA IS IT ALWAYS WITH VON HIPPEL LINDAU SYNDROME? A RARE CASE OF LITERATURE

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INTRODUCTION: Hemangioblastoma (HB) is a vascular tumor of the CNS, accounting for only 1-2.5% of all intracranial tumors and approximately 10% of all posterior fossa tumors.¹ It occurs most often in the cerebellum.^{2,1}

Haemangioblastomas occur both in patients with Von Hippel Lindau disease (VHL) as well as sporadically. About 45% of those with VHL develop HBs and 20% of those with HB have VHL.¹ Here we discuss a rare case of HB of brain associated with VHL in a young female.

CASE REPORT: A 38 yrs old female presented to our institute with history of headache, vomiting and gait abnormality for 2 months. She gave a history of brain tumor for which she underwent surgery three times since 2013.

Initially in May 2013 she had similar complaints and CT scan of brain showed heterogenous density (4.8x3.4 cm) in the left cerebellar hemisphere (LCH) causing obstruction of the 4th ventricle (FV) and dilatation of the supratentorial ventricular system (STVS) suggestive of posterior fossa space occupying lesion (SOL). On the MRI of brain the lesion was hyperintense on T2 WI, hypointense on T1 WI FLAIR with isointensity (1.4x1.2cm) noted within pial surface (mural nodule) with perilesional edema.

She had a left sub occipital craniotomy and excision of the SOL. Cytology from the SOL (Squash) was suggestive of HB and histopathological evaluation (HPE) was suggestive of a vascular lesion.

Patient did not receive any Adjuvant treatment and the CT scan brain at 1 month follow-up showed post-operative changes.

One year later patient was symptomatic and MRI brain showed a recurrent lesion (4.8x 4.2 cm) in the LCH. She underwent excision biopsy of the SOL. The HPE showed HB. Patient was treated symptomatically and kept on follow-up.

Four months later in September 2014 patient was symptomatic and on examination had bilaterally decreased visual acuity and papilledema. CT scan brain showed a 4.6x3.6 cm lesion posterior to the FV with compression of FV and dilatation of the STVS. Then patient had a right Ventriculo Peritoneal (VP) shunt placement. A Cranio Spinal Fluid analysis was also done which was within normal limits.

Patient was referred to our institute for further management of the recurrent Cerebellar HB in November 2014. When she presented to us, she was wheel chair bound accompanied by a family member. On examination she had intention tremor, horizontal nystagmus, ataxic gait and left hemiparesis.

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Patient is a known type-2 diabetic and hypertensive on oral medications for 7 years. She has three children with last child born 17 years ago. Her mother had history of vision loss at the age of 65 years for unknown reason but no history of brain or kidney tumors in the family.

As patient was having HB of brain which is a rare tumor, we reviewed the literature and found the association of HB to VHL. We investigate the patient for retinal and other visceral lesions.

She underwent CT scan of abdomen and pelvis showing bilateral enlarged kidneys with bosselated contours, multiple cysts of varying sizes (largest 4x3.4cm). Also multiple cysts were noted in the pancreas (largest 1.4x1.5 cm) suggestive of VHL.

Figure 1a: Axial image of the CT scan abdomen and pelvis showing bilateral enlarged kidneys with multiple cysts.

Fig. 1a

Figure 1b: Axial image of the CT scan abdomen and pelvis showing multiple cysts in the pancreas.

Fig. 1b

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As VHL patients could have spinal lesions as well, patient had a CT scan and MRI of the spine along with MRI scan of the brain. There was a large hypervascular enhancing mass lesion (4.5x4.7x4.5cm) in the vermis and the LCH with significant perilesional edema extending into brainstem with mass effect over the FV s/o recurrence. Few well defined nodular lesions were noted in the right cerebellar hemisphere with minimal perilesional edema s/o small HBs.

Figure 2a: Axial images of CT scan brain showing mass lesion in the vermis and the LCH with perilesional edema.

Fig. 2a

Figure 2b: Coronal images of CT brain showing mass lesion in the vermis and the LCH.

Fig. 2b

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Cervicomedullary junction showed a cystic mass lesion 2.4x1.4 cm with mural nodule in the inferior aspect measuring 9x6 mm with significant compression over the cervicomedullary junction in the region of foramen magnum. Spinal cord from C2 to C5 showed cord edema. Multiple renal cortical cysts were noted in both the kidneys. Chest x-ray showed VP shunt and no active lung lesions.

Figure 2c: Coronal images of the MRI brain and spine showing mass lesion in the vermis and the LCH. Also multiple renal cortical cysts in both the kidneys.

Fig. 2c

Ophthalmic examination showed a normal anterior segment but fundus exam of right eye showed a Hemangioma, while the left eye exam was normal.

Figure 3a: Fundus photo of the right eye showing posterior pole with dilated tortuous inferotemporal retinal vein.

Fig. 3a

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Figure 3b: Fundus photo of right eye showing retinal angioma at the extreme inferior temporal periphery of the vein.

Fig. 3b

Figure 3c: Left eye Fundus showing normal features.

Fig. 3c

As clinical examination and investigations were suggestive of VHL, patient underwent chromosomal analysis for VHL gene. However chromosomal analysis was normal with 46XX karyotype.

Fig. 4: Chromosomal analysis showing 46XX

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Patient labs including complete blood picture, renal and liver function tests were within normal limits.

Patient was having recurrent HB of the cerebellum and the tumor was inoperable. So we planned External Beam Radio Therapy (EBRT) for the cerebellar and the cervicomedullary lesions with Photons on the Linear Accelerator by the Intensity Modulated Radio therapy (IMRT). She received a total EBRT dose of 54 Gray in 27 fractions from 1st December 2014 to 6th January 2014. IMRT plan was done with 6 fields using the Eclipse 8.9 treatment planning system. Organs at Risk (OAR) constraints were achieved for brainstem, spinal cord, optic chiasm, pituitary and right and left orbits, optic nerve, lens and cochlea. During Radiotherapy patient had vomiting which was controlled with symptomatic treatment.

Figure 5a: Planning images of the EBRT plan (IMRT), with Photons on the Linear Accelerator.

Fig. 5a

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Figure 5b: Dose Volume Histogram images for the planning target volume (PTV) in red and OARs.

Fig. 5b

The right retinal hemangioma was treated with focal Photocoagulation in November 2014 and again in January 2015.

During first follow-up in Feb-2015, at 1 month post-EBRT, patient was symptomatically and neurologically better. She was able to ambulate with support.

DISCUSSION: In patients with VHL, HB of brain are usually multiple along with retinal angiomas, renal cell carcinoma, pheochromocytomas, pancreatic cysts and neuroendocrine tumors. Overview by specific benign and malignant lesions is sufficient to make the diagnosis. If no known family history exists, at least 2 cerebellar HBs or 1 HB plus 1 visceral tumor are necessary to justify the diagnosis of VHL.^{2,1,3}

Hemangioblastoma appears to be associated with VHL and all patients with sporadic nonhereditary tumors should be evaluated for evidence of VHL.³ Sporadic cases make up approximately 75-80%. Single tumors may be sporadic, but multiple tumors are almost always associated with VHL. Sporadic tumors appear in the 5th and 6th decade of life, whereas VHL-associated tumors manifest in the 3rd or 4th decade. About 45% of those with VHL develop HBs and 20% with HB have VHL.^{2,1}

They are WHO grade I tumors that can occur in the central nervous system or kidneys, liver and pancreas.¹ The nidus of the tumor abuts the pia mater, from which the tumor receives its vascular supply. The tumor is more frequently superficial than deep.³

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Location of the HBs is as below:

- Intracranial: 87-97%.
 - 95% in posterior fossa:
 - 85% in cerebellar hemisphere.
 - 10% in the cerebellar vermis.
 - 5% medulla.
 - Only rarely do they extend beyond the cerebellum into the cerebellopontine angle.
 - 5% supratentorially (typically in the optic radiations).
 - Cerebral HBs are only really seen in patients with VHL.
- Spinal: 3-13%.

Symptoms and signs included headache (75%), ataxia (55%), dysmetria (29%), and hydrocephalus (28%).²

Contrast enhanced MRI is the best method for screening patients with VHL and the first evaluation used in symptomatic patients. Preoperative angiography is important for defining feeding vessels and aiding in embolization.^{2,3}

The optimum contemporary management of cerebellar HBs in VHL has not been defined. Surgical resection is usually curative and large lesions are made easier by preoperative embolisation. Tumor recurrence is avoided by meticulous extracapsular resection.⁴ Adjuvant radiotherapy is used in patients with incomplete resections. Recurrence can be seen in up to 25% of patients.¹

Differential diagnosis of HB of brain includes benign, malignant tumors, AVMs and infarcts.¹

Patients with VHL should be screened and follow-up studies should be performed regularly for renal, adrenal, liver and pancreatic tumors. Early treatment improves the outcome. Using MRI and CT at 1- to 2-year intervals, Conway et al identified 74% of lesions that required surgery before the patients became symptomatic.^{2,3}

CONCLUSION: HB of brain is a rare tumor sometimes associated with VHL. Patients with HBs need to be investigated for tumors at other sites and if positive they need genetic testing and chromosomal analysis to rule out familial syndrome. Regular follow up of the patients by clinical examination and imaging is also warranted to detect new lesions.

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