Misdiagnosed Cavernous Malformations of Brain

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Introduction

Arteriovenous malformations, cavernous malformations (CMs), venous angiomas, and capillary telangiectasias are four main central nervous system (CNS) vascular malformations. Of these CMs are found in 0.5-0.7% of the general population. CMs are composed of low flow abnormal vessels and are of familial or sporadic type. Familial CMs are autosomal dominant transmitted and are associated with CCM-1, CCM-2 and CCM-3 genes. Supra tentorial brain CMs are common as compared to the infra tentorial. Most of the CNS CMs remain stable and asymptomatic. Occasionally these grow and are complicated by recurrent haemorrhages. CMs located in brain stem have 30 times higher risk of hemorrhage compared to other locations. A number of clinical scenarios can complicate CNS CMs. Subarachnoid or intraparenchymatous haemorrhages, seizures, and fluctuating or slowly progressive neurologic deficit are the listed complications. This case report describes a patient with CMs who was misdiagnosed as neoplastic lesion of brain.

Case Summary

A 30 years old epileptic female presented with typical generalized tonic clonic fits for 10 hours. Tongue bite and urinary incontinence were also noted. She was managed on lines of status epilepticus. At admission and subsequent examinations showed that she had nystagmus, and right sided upper motor neuron type hemiparesis. MRI brain (Figure 1 and 2) showed 16x13 mm, contrast enhanced hemorrhagic lesion in the right frontal region. Hemorrhagic lesions were also noted in left cerebral peduncle extending into superior part of pons, left half of medulla anteriorly, and left parietal region.

Three years back she was labeled as a probable case of brainstem lymphoma when she had sudden onset of headache and right sided weakness. CT scan and MRI of brain showed slightly contrast enhancing, multiple areas of hemorrhage involving brainstem and right frontal lobe (Figure 3 and 4). Considering the risks of neurosurgical intervention, patient and family decided against neurosurgical intervention. Patient was discharged on anti epileptic medication.

Comparison of the previous and current MRI brain showed that right frontal lesion was unchanged, left cerebral peduncle lesion had decreased in size, medullary lesion was inconspicuous in previous study, and previously seen hemorrhagic lesion in the left parietal region had regressed. Based on the presence of multiple hemorrhagic areas which had either decreased in size or regressed, diagnosis of multiple hemorrhagic CMs of the brain was made. The patient is currently better after modification of anti epileptic medications, general care and physiotherapy. She has been asked to come for regular follow up and importance of serial brain imaging has been explained.

Discussion

Figure 1: Fresh MRI showing the right frontal hemorrhagic lesion.
Figure 2: Fresh MRI showing regression of old brain stem hemorrhage.
Figure 3: Three years old MRI brain of the patient showing right frontal region hemorrhage.
Figure 4: Three years Old MRI Brain showing brain stem hemorrhage.
Discussion
Intracranial CMs consist of thin walled sinusoids. These are present at birth. Although these do not have a capsule but are separable from brain parenchyma due to presence of reactive gliosis membrane. These features make CMs surgically resectable. Size of CMs varies from few millimetres to centimetres. Although these can be found anywhere in CNS, 70-80% of CMs are present in supra-tentorial locations. Pathologically, CMs are benign but can expand due to repeated haemorrhages and clinical features may develop as a consequence. CMs may become calcified as well. Site, size, and haemorrhage in the CMs are determinative of clinical features. Progressive focal neurological deficits, fits, and acute onset headache are frequent presenting features of CMs. Serial monitoring of the patients with CMs is required. Anti epileptic medications are prescribed for fits. Family screening has to be done wherever considered appropriate.

References