AN INTERESTING CASE OF CAVERNOMA

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ABSTRACT:

14-year-old boy was admitted with sudden weakness of right side of upper limb and lower limb with right sided facial nerve palsy. He had no history of fever, trauma, and seizures. MRI brain with contrast showed multiple well defined intracranial predominantly hypointense lesions with edema in lesion involving left hemi pons possibly representing cavernomas. Child managed conservatively improved symptomatically

KEYWORDS: Cavernoma, Case, MRI, Haemorrhages


INTRODUCTION:

Cavernoma is an intracranial cerebral vascular malformation. The term cavernoma is derived from the word cavern (large cave) the abnormal malformation in the capillary have defective walls with lack of surrounding support hence they are easily damaged and they are susceptible to bleed, so the capillaries easily leak and are filled with blood creating a cavern. After bleeding resolved they do not return their normal state leaving behind empty space known as caverns. There are various terms for cavernoma such as cavernous angioma, cavernous hemangiomas or cerebral cavernous malformation. According to ISSVA (International Society for the Study of Vascular Anomalies) classification cavernoma now classified under slow flow vascular malformation. Most
common symptoms associated with paralysis, headache, seizures, hearing and visual disturbances. Most severe complication is hemorrhage. It can be sporadic and familial. In familial inheritance will be autosomal dominant pattern involving mutation in the \( \text{KRIT1} \) (CCM1), \( \text{CCM2} \), and \( \text{PDCD10} \) (CCM3) genes. Most commonly used investigation for diagnosis is MRI however CT and Angiography can also be used. Genetic testing can be used to identify the familial cases. Our case is a 14 year old child with familial cavernous malformation with neurological deficits and family history of cavernoma in elder brother who is asymptomatic.

**CASE REPORT:**

14 year old boy came to our paediatric outpatient department with history of headache for 3 days- initially over the forehead and later involved occipital region, continuous, dull aching pain, relieved by paracetamol. He complaints of weakness of right upper limb and lower limb, which was gradual in onset and progressive in nature. H/o difficulty in dressing and eating but was able to walk with difficulty. No history of difficulty in speech. No behavioural abnormalities. H/o double vision. No h/o fever within this month. No h/o loss of consciousness/giddiness/truma/seizure/vomiting. No h/o bladder or bowel incontinence. No h/o nasal regurgitation of feeds/deviation of angle of mouth. No h/o loss of pain, touch, temperature. No h/o Tuberculosis, Bronchial Asthma. No h/o similar illness in the past. No h/o any hospitalisation. Second born of a third degree consanguineous marriage. First sibling is also a known case of familial cerebral cavernous malformation and not on any treatment and has no neurological deficit follow up with yearly MRI.

On presentation patient was conscious and oriented. No neurocutaneous markers. He had weakness of right upper and lower limb of power 4/5 with no involvement of cranial nerves, which gradually progressed with involvement of cranial nerves and progressive weakness and exaggerated DTR. First affecting right 7th CN with deviation of angle of mouth to left and loss of nasolabial fold on right side. then cranial nerves 6th, 11th, 12th on right side with extraocular movements restricted on left side. Diplopia + .Left sided lateral rectus palsy, convergent squint, Shrugging of shoulders decreased on right side, Difficult turning of head to left side, tongue deviated to right side. Weakness progressed on right upper and lower limb to grade 2 power with exaggerated DTR and extensor plantar reflex on right side and also involving left upper and lower limb power of grade3, exaggerated DTR and equivocal plantar. Sensory system was not affected. No signs of raised ICP/ meningeal irritation. Hence the neurological examination revealed bipyramidal weakness and UMN right facial nerve weakness with involvement of cranial nerves right 6,11 and 12.

Baseline Investigations were done and within normal limits. MRI brain with contrast showed multiple, well defined intracranial predominantly hypointense lesions with edema in lesion involving left hemipons possibly representing cavernomas. Both Neurologist and neurosurgery examined the patient and advised for conservative management. Child was treated conservatively with IV hypertonic saline, Inj. Levipil, Inj. Dexa. Ophthalmology opinion was taken and revealed bilateral 6th nerve involvement. CT brain showed same as MRI report. In view of aggravating signs (involvement of 12th nerve) interventional radiologist opinion obtained and they advised conservative management. Patient was started on Intensive physiotherapy. Patient improved symptomatically and power in limbs is gradually improving. Patient is being discharged with home physiotherapy.
DISCUSSION:

Cavernoma are cerebral venous malformation. It is one of the most common cerebral vascular malformations. Other common cerebral vascular malformations are capillary telangiectasia, developmental venous anomaly, vein of galen malformation, mixed vascular malformation. According to ISSVA (International Society for the Study of Vascular Anomalies) classification of vascular anomalies cavernoma is termed under slow flow venous malformations. Classification for cerebral cavernous malformation is ZABRAMSKI classification.

EPIDEMIOLOGY: Both males and females are affected equally with no sex predominance. The incidence is widely variable and prevalence in the general population is 0.02 to 0.13%. Cavernous malformations, accounting for approximately 5-15% of all vascular abnormalities in the central nervous system. It can occur sporadically and hereditary. Familial cavernoma in Hispanic patients are more when compared to Caucasians.
PATHOLOGY: Cavernoma is a benign condition. It can occur as single or multiple lesions. Multiple lesions are commonly seen with familial cavernomas. Cavernomas are well circumscribed multilobulated berry like lesion. Size of cavernoma ranges from few millimetres to centimetre. It more frequently occurs subtentorially. Cavernomas are found to occur both cortically and deep (thalamus, basal ganglion, cerebellum brainstem). Rarely lesions were found in cerebropontine angle, pituitary and intraventricular areas. Often associated with developmental venous anomaly forming mixed vascular malformation. Blood vessels of cavernoma are more prone for leakage as their walls are hyalinized and defective smooth muscle. Abnormal vasculature can lead to venous congestion, thrombosis, phlebolith formation with gradual expansion of lesions. Three genes CCM1, CCM2, and CCM3 involved in angiogenesis and structural integrity of endothelial cells. Abnormality of these genes can lead to cavernomas.

CLINICAL FEATURES: Cavernoma mostly be asymptomatic and were found incidentally. Most often diagnosis is made when MRI is done after any focal neurological deficit or seizure like activity. It can occur in any age. Symptoms and severity will depend on location of the lesion. Typical symptoms will be seizures, double vision, headache, weakness of arms and legs, memory loss, hydrocephalus. Small bleeds usually do not cause any symptoms.

COMPLICATIONS: Recurrent bleeding with progressive neurological deficit. Person with history of previous bleed are more risk for recurrent haemorrhage.

INVESTIGATION: Most of cavernous malformation found in subtentorial area can occur anywhere including brainstem, spinal cord. They can be single as well as multiple. Multiple cavernoma mostly seen with familial cases.

CONVENTIONAL MRI: main modality which accurately demonstrate the cavernous malformation “popcorn or berry” appearance surrounded by a rim of hemosiderin deposits

Zabramski classification of cerebral cavernomas based on MRI

- Type I: subacute haemorrhage
  - T1: hyperintense
  - T2: hypo or hyperintense

- Type II: most common type - classic "popcorn" lesion
  - T1: mixed signal intensity centrally
  - T2: mixed signal intensity centrally
  - low signal rim with blooming on T2* sequences

- Type III: chronic haemorrhage
  - T1: hypointense to isointense centrally
  - T2: hypointense centrally
  - low signal rim with blooming on T2* sequences

• Type IV: multiple punctate microhaemorrhages
  o T1: difficult to identify
  o T2: difficult to identify
  o T2* Gradient Echo: "black dots” with blooming
  o difficult to distinguish from small capillary telangiectasias

GRADIENT RECALLED ECHO (GRE) MR IMAGING: lesion will be prominent bloom (clearly visualized). Magnetic susceptibility effect due to hemosiderin-filled brain tissue which shows very distinct hypointensity. It is useful for detecting smaller lesion which may be missed by conventional MRI. It accurately identifies all foci and size of lesion.

SUSCEPTIBILITY-WEIGHTED MR IMAGING: More sensitive method to detect small cavernoma that cannot be found by conventional method. They accurately detect deoxyhaemoglobin and hemosiderin

DIFFUSION TENSOR (DT) IMAGING: Used intraoperatively for visualization of lesion and surrounding areas. DT helps to visualize the tracts (the corticospinal tract, the medial lemniscus and the cranial nerve nuclei) and prevent its damage. Studies have shown use of diffusion tensor imaging reduced the occurrence of seizure and other morbidity post operatively.

COMPUTED TOMOGRAPHY: CT will identify only large lesions unless there is bleed and calcification. High density, well defined lesion with or without calcification. CT may lead to under diagnosis of the disease.

Angiography (DSA): Angiography is done to rule out arteriovenous malformation. Cavernoma is an occult malformation angiographically. It is used to demonstrate association of developmental venous anomaly with cavernoma. Other imaging technique is needed for the diagnosis.

TREATMENT: Management of cavernoma depends on many factors like asymptomatic, seizure, bleeding, number of cavernoma, location, associated with developmental venous anomaly. For asymptomatic patients no need for therapy regular follow-up with MRI scans is advised. Due to the potential risk of intervention conservative management is preferred like seizure were managed with antiepileptic drugs. For symptomatic cavernoma like uncontrolled seizure, hemorrhage with focal neurological deficit surgery is considered. Surgery is the only cure for cavernoma. In each patient the risk is different which depend on size, number, location. Superficial and subtentorial cavernomas are easily approached. Brainstem cavernomas are at high risk with operation. Microsurgical resection is done by small craniotomy and with the help diffusion tensor imaging to avoid damage to important tracts. Constant micro electrical neuromonitoring is done during procedure which alerts the operator about the sensitive structures. For any neurological deficit due to cavernoma postsurgical rehabilitation is recommended.

CONCLUSION: A space occupying lesion with sudden onset of focal neurological deficit. MRI brain is a sensitive investigation. Microsurgical resection is considered for symptomatic patients.

REFERENCES