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Rare Case of Cva – Possible Phace Syndrome

Medical Science

KEYWORDS :

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1. Introduction

cavernous angiomas (cavernomas).these are of unknown aetiology,. Cavernomas are predominantly supratentorial, most commonly affecting the temporal, frontal and parietal lobes. The most frequent infratentorial site is the pons. Cavernomas may present with seizures, intra-cerebral haemorrhage or progressive neurological deficit. The last of these is more likely to occur in brainstem cavernoma. The majority of cavernous angiomas are asymptomatic and are frequently found on MRI performed for other reasons. [1] we are here with presenting a case report of cavernous angioma and hypo plastic vertebral artery presenting with neurological deficit.

2.CASE REPORT

A 40 year male patient presented with Difficulty in walking since 8 days, Slurring of speech since 8 days, Dizziness since 8 days.

Patient was apparently asymptomatic 8 days back. Then he developed sudden onset difficulty in walking with swaying towards right side. H/o difficulty in taking food to mouth was present. H/o headache and vomiting present. H/o slurring of speech was present. ,H/o dizziness was present, No difficulty in understanding the speech. No positional changes. Not associated with ear discharge and deafness.

No h/ofever, loss of consciousness, seizures, trauma, bowel or bladder involvement and recent vaccination.

Past History

H/o Cerebrovascular accident 2 times. (4 years and 6 months ago ago, recoverd). Hypertensive since 4 years on regular treatment.Diabetic since 6 months on regular treatment.

Family History

Elder sister died of CVA at the age of 50 years.

Personal History

Takes mixed diet, Normal appetite, Normal bowel bladder function, Normal sleep, Non-alcoholic, Non-smoker.

General Examination

Patient is conscious, coherent, No pallor, icterus, cyanosis, clubbing, pedal edema, lymphadenopathy, No neurocutaneous markers, PR: 86/ min, BP: 150/90 mm of hg, Temp: normal,RR: 18/ min.

Nervous system examination

Mental functions normal, cranial nerves: left 5 th, 6 th(Fig 1) LMN type of 7th, 8 th nerve palsies present.Horizontal nystagmus present and other cranial nerves are normal

Motor system: Power 4/5 on right side. Hypotonia on right side Normal power and tone on left side**.**

Superficial and deep tendon reflexes are normal, Incoordination present on right side, Sensory system : normal, Signs of cerebellar dysfunction present on right side, no signs of meningeal irritation, skull and spine normal,Other systems : normal.

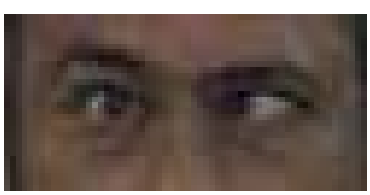

Fig.1: Left lateral rectus palsy, Left LMN facial palsy.

Investigations

C.B.P, C.U.E, blood urea, serum creatinine, lipid profile normal, R.B.S: 204 mg/dl, Computerised Tomography of Brain: normal study, 2D ECHO: concentric LVH present, Carotid Doppler: normal, serum homocysteine, protein c,protein s, levels were normal, ANA profile was normal, M.R.I brain(Fig 2): periventricular small vessel ischemic changes.-cerebellar atrophy/hypoplasia noted - altered signal intensity hypointense on T1W1,T2W1 noted in the lower pons without any perilesional edema, M.R.I. brain contrast study(Fig 3) s/o cavernous angioma, C.T.Angiogram(Fig 4): s/o hypoplastic right vertebral artery.

Final diagnosis

Case of recurrent young stroke with cavernous angioma in the pons, cerebellar hypoplasia/atrophy and hypoplastic right vertebral artery – Possible PHACE Syndrome.

Fig.2:MRI Brain showing hypo intense lesion in lower pons without any perilesional oedema

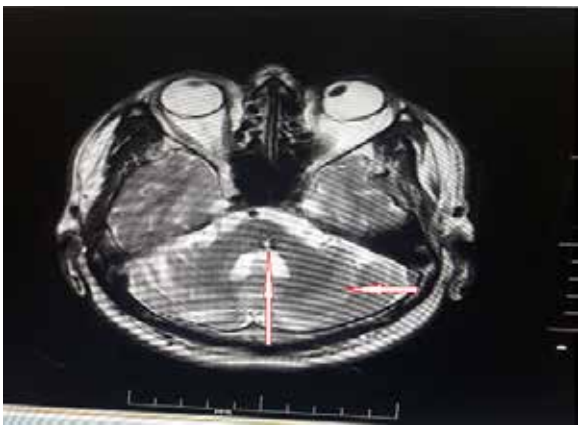

Fig.3 : MRI Brain with Contrast showing prominent folia in Cerebellum s/o Cerebellar atrophy/hypoplasia, and cavernous angioma in pons

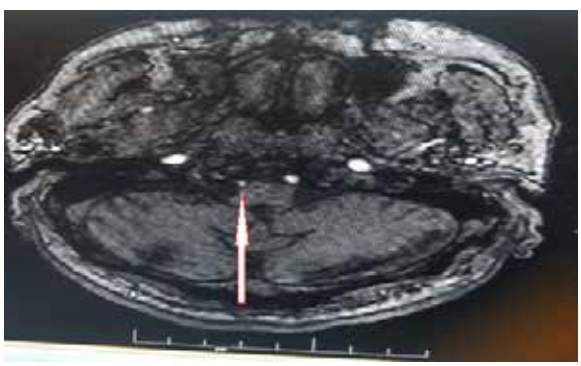

Fig. 4: Showing Hypoplastic Right vertebral artery

DISCUSSION

What is PHACE syndrome?

A syndrome is defined as a recognizable pattern of medical conditions that occur together. **PHACE syndrome** [2] is a disorder characterized by large infantile hemangiomas of the face, scalp and neck associated with developmental defects of the brain, blood vessels, eyes, heart and chest wall.

History of PHACE syndrome

PHACE syndrome [2] is a relatively newly discovered entity and was first described in 1996 by an astute physician who recognized a pattern of problems in babies with large facial hemangiomas**.**

Dr. Ilona Frieden and colleagues published a medical paper detailing 5 babies with large facial hemangiomas and anomalies of the brain, aortic arch, eye and arteries.

Frieden created the term **PHACE**, which refers to ***P***osterior fossa anomalies, ***H***emangioma, ***A***rterial lesions, ***C***ardiac abnormalities/***C***oarctation of the aorta, and ***E***ye anomalies.

Every child diagnosed with PHACE syndrome [3] has a different combination of abnormalities associated with the syndrome.

Not every affected child has all of the same symptoms, and there is a spectrum of severity. In most cases, there is no history of PHACE syndrome or similar medical conditions in the family.

For reasons we do not understand, girls are more likely to get PHACE syndrome when compared to boys, but the severity of the disease does not seem to be worse in either sex. At this point, little is known about what problems may occur as children with PHACE syndrome get older. The cause of PHACE syndrome remains unknown. However, nationwide research efforts are beginning to provide valuable information about PHACE syndrome with the goal of improving clinical care for these children.

How common is PHACE syndrome?

Although relatively uncommon, more than 400 cases of PHACE syndrome [4] have been reported in the medical literature.

This number, however, is most likely an underrepresentation of the true number of PHACE syndrome cases due to a variety of reasons.

First, the reported cases found in the literature often have the most severe abnormalities associated with PHACE syndrome. Thus, children who have a more mild form of PHACE syndrome may not be reported in medical journals. Second, some children that were previously misdiagnosed with other diseases, most notably Sturge-Weber Syndrome, may actually have PHACE syndrome. As understanding and recognition of PHACE syndrome increases, the reported number of patients diagnosed with the syndrome is expected to increase.

How is PHACE syndrome diagnosed?

The identification of PHACE syndrome [2-4] is made by a clinical diagnosis. This means there is no one sign or symptom that will indicate a diagnosis of PHACE syndrome. Currently, there is no blood test or genetic test for PHACE syndrome. Physicians use a combination of signs and symptoms called diagnostic criteria to make the diagnosis.

Most children with PHACE syndrome have a large hemangioma on the face, scalp or neck. One study found that 31 percent of children with large infantile hemangiomas on the face or scalp have PHACE syndrome. Rarely, children with large hemangiomas of the arm and/or trunk may also have PHACE syndrome. Children with a large segmental hemangioma of the head and neck should undergo a thorough evaluation, especially of the brain, heart, blood vessels and eyes.

What are the diagnostic criteria for PHACE syndrome?

The diagnosis of PHACE syndrome [5] is made by using a combination of major and minor criteria. Major criteria are findings that are common in PHACE syndrome but rare in other medical conditions. Minor criteria are findings that are seen in PHACE syndrome but can also be found in other conditions.

Affected children are classified into two categories, definite PHACE syndrome and possible PHACE syndrome, based on the nature and number of criteria met.

Definite PHACE syndrome requires the presence of a characteristic segmental hemangioma greater than 5 cm in diameter on the face (or scalp) plus one of the major criterion or two minor criteria.

Possible PHACE syndrome can be diagnosed in one of three different combinations:

a) Facial hemangioma greater than 5 cm in diameter plus 1 minor criterion; b) hemangioma of the neck or upper torso plus 1 major criterion or 2 minor criteria; orc) no hemangioma plus 2 major criteria.

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Organ system	Major criteria	Minor criteria
Cerebrovascular	Anomaly of major cerebral arteries Dysplasia of the large cerebral arteries Arterial stenosis or occlusion with or without moyamoya collaterals **Absence or moderate - severe hypoplasia of the large cerebral arteries** Aberrant origin or course of the large cerebral arteries Persistent trigeminal artery Saccular aneurysms of any cerebral arteries	Persistent embryonic artery other than trigeminal artery Proatlantal intersegmental artery (types 1 and 2) Primitive hypoglossal artery Primitive otic artery
Structural Brain	Posterior fossa anomaly Dandy-Walker complex or **unilateral/bilateral cerebellar hypoplasia/dysplasia**	**Enhancing extra-axial lesion with features consistent with intracranial hemangioma** Midline anomaly Neuronal migration disorder
Cardiovascular	Aortic arch anomaly Coarctation of aorta Dysplasia Aneurysm Aberrant origin of the subclavian artery with or without a vascular ring	Ventricular septal defect Right aortic arch (double aortic arch)
Ocular	Posterior segment abnormality Persistent fetal vasculature (Persistent hyperplastic primary vitreous) Retinal vascular anomalies Morning glory disc anomaly Optic nerve hypoplasia Peripapillary staphyloma Coloboma	Anterior segment abnormality Sclerocornea Cataract Coloboma Microphthalmia
Ventral or Midline	Sternal defect Sternal cleft Supraumbilical raphe	Hypopituitarism Ectopic thyroid

Definite PHACE syndrome		
Facial hemangioma greater than 5 cm PLUS 1 major OR 2 minor criteria		
Possible PHACE syndrome		
Facial hemangioma greater than 5 cm PLUS 1 minor criteria	Hemangioma of the neck or upper torso PLUS 1 major OR 2 minor criteria	**2 major criteria without hemangioma**

The patient has presented with 2 major criteria (Hypoplasia of Right vertebral artery and Hypoplasia /Aplasia of Cerebellum with out Facial Haemangioma) considering POSSIBLE PHACE SYNDROME .

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