Case Report

Basilar invagination presenting as recurrent posterior circulation stroke with locked-in syndrome in a child: a case report

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ABSTRACT

Basilar invagination is a type of craniocervical junction anomaly which occurs when the tip of odontoid process migrates upward into the intracranial space through foramen magnum and most commonly present as progressive neurological deficit implicating the high cervical cord, lower brainstem and cranial nerves. It can rarely present as vertebro-basilar territory insufficiency and occlusion, leading to stroke. Treatment is typically surgical and continues to centre on varying combinations of anterior or posterior decompression with or without traction or fusion. We present a 13-year-old boy with basilar invagination diagnosed on MRI with angiogram, which was missed on initial presentation, leading to recurrent posterior circulation stroke progressed to Locked-in syndrome. This case report emphasis the need of screening for craniovertebral junction anomalies in young patients presenting with posterior circulation stroke.

Keywords: Basilar invagination, Craniovertebral junction anomalies, Locked-in syndrome, Posterior circulation stroke

INTRODUCTION

Congenital cranio-vertebral junction (CVJ) anomalies encompass the developmental defects of the atlas and axis vertebrae along with occipital bone surrounding the foramen magnum. CVJ developed from the mesodermal somites to form four occipital and two cervical sclerotomes, during fetal development. Defects occurring in the third and fourth week of gestation during embryogenesis can cause CVJ anomalies which may implicate either the neural or skeletal structures or both.1,2

Basilar invagination/impression (BI) refers to CVJ anomaly of occipital bone and upper cervical spine, characterized by upward protrusion of the odontoid process into the intracranial space. It is a rare clinical condition which may lead to static or dynamic stenosis of foramen magnum and obstructive hydrocephalus or compression of medulla oblongata and sudden death.1-3

"Locked-in syndrome" refer to a neurological condition manifested as quadriplegia, lower cranial nerve paralysis, and mutism with preserved consciousness via intact vertical eye movements by which patient is able to communicate intelligibly using eye blinking. Most cases of locked-in syndrome are caused by basilar artery occlusion resulting in brainstem infarction in the ventral pons.

Vertebo-basilar circulation insufficiency (VBI) and occlusion leading to posterior circulation stroke (PCS) are rare presentation of BI.1,3 We report a case of 13-year-old boy with BI, having recurrent PCS progressed to locked-in syndrome.
CASE REPORT

A 13-year-old boy was admitted with complaints of sudden onset headache, vomiting, vertigo and paresthesia in left half of the body followed by unable to speak and move his body for 5 days.

Perinatal history was uneventful with normal age appropriate developmental milestones. Five years back, patient was hospitalized for headache, vomiting, and fluctuating sensorium having aphasia, left lateral rectus palsy, and left sided spastic hemiparesis. CSF examination, abdominal ultrasound, echocardiography and color Doppler study of neck vessels were within normal limits. Axial unenhanced CT showed acute ischemic lesions/infarctions in bilateral thalami and right cerebellar hemisphere. Attendants refused for MRI with angiography. Patient was treated conservatively and residual neurodeficit persisted in form of left lateral rectus palsy and minimal left side weakness.

Figure 1: MRI shows acute infarcts in right thalamus, pons and right cerebellar hemisphere: (a) axial fluid-attenuated inversion recovery images show areas with increased signal intensity. (b) Diffusion weighted MR images; (b 1000 sec/mm²) show increased signal intensity and (c) ADC maps; Decreased ADC values in the same areas due to the restricted mobility of water molecules.

Figure 2: Non-contrast Magnetic resonance angiography; Non-visualization of bilateral intracranial vertebral arteries at the level of foramen magnum, entire length of basilar artery and bilateral anterior cerebellar arteries likely as a result of changes in the craniovertebral anatomy; attenuation of bilateral posterior cerebral arteries was noted.

Figure 3: Sagittal T2-weighted Magnetic resonance imaging; Basilar invagination as tip of the odontoid process projecting above the foramen magnum, incomplete atlanto-axial assimilation, reduction of the anterior subarachnoid space and compression over medulla oblongata and upper cervical cord.

Presently, there was no history of fever, rash, ear discharge, seizures, head/neck/back trauma, visual disturbances and discoloured urine. Vital parameters were within normal limits with no pallor, icterus, cyanosis, lymphadenoapathy, clubbing, facial dysmorphism and neurocutaneous stigmata. Glasgow coma scale score was 6/15. Patient was in Locked-in syndrome with quadriplegia, mute, preserved eye movements, and without complete loss of consciousness. Pupils were 3 mm with normal direct and indirect light reflex bilaterally. Horizontal gaze nystagmus in right eye with fast component towards right was present. Facial weakness and pooling of oral secretions were present. Patient had hypotonia, brisk deep tendon reflexes, absent abdominal and cremasteric reflex and bilateral extensor plantars. Sensory system and cerebellar signs can’t be assessed. There were no signs of meningeal irritation. Skull and spine were normal. A review of other systems was within normal limits.
Investigations revealed a normal hemogram, ESR, blood sugar, electrolytes, cholesterol, triglycerides, coagulation profile, ECG, chest X-ray, liver and kidney function tests. Tests for rheumatoid factor, ANA, ASO titre, HIV, HBsAg, Widal and Mantoux were negative.

Non-contrast MRI with angiography revealed fresh infarcts in right thalamus,pons and right cerebellar hemisphere in posterior circulation territory. Sagittal T2-weighted images of CVJ showed basilar invagination as tip of odontoid process projecting above the foramen magnum, incomplete atlanto-axial assimilation, reduction of the anterior subarachnoid space, compression over medulla oblongata and upper cervical cord. Non-contrast Time of flight MRA confirmed occlusion of bilateral intracranial vertebral arteries at the level of foramen magnum and entire length of basilar artery with non-visualization of bilateral anterior cerebellar arteries and attenuation of bilateral posterior cerebral arteries.

Bilateral vertebral arteries proximal to the level of foramen magnum, bilateral intracranial internal carotid, middle cerebral and anterior cerebral arteries were of normal calibre. Chronic lacunar infarct was seen in left centrum semiovale with small gliosis in left thalamus.

Patient referred for neurosurgical intervention, but attendants refused and left our hospital without further follow-up.

**DISCUSSION**

BI occurs when tip of odontoid process migrates upward into the intracranial space through foramen magnum. It occurs congenitally, in bone diseases and medical conditions (e.g. Chiari malformation, Down syndrome, Klippel Feil, osteogenesis imperfecta, and rheumatoid arthritis), due to ligamentous laxity and trauma.\(^1\)\(^2\)\(^3\)\(^4\) Atlanto-axial dislocation (AAD), occipitalisation of atlas, and fusion of C2-C3 vertebrae are the commonest CVJ anomalies occurring in combination in India, whereas BI is more common in the West. Mean age of manifestation of congenital CVJ anomalies is 25 years, and males outnumber females.\(^1\)\(^2\)\(^5\)

CVJ anomalies most commonly present as progressive neurological deficit (76-94%) implicating the high cervical cord, lower brainstem and cranial nerves. VBI and/or PCS are rare presentation attributed to adequacy of the circulation via two vertebral arteries and circle of Willis.\(^1\)\(^2\)\(^5\) Commonest CVJ anomalies causing stroke/VBI is AAD followed by odontoid aplasia, BI, occipitalisation of the atlas, Klippel-Feil anomaly and anomalous occipital bone.\(^5\)

In one study, only 11% of the VBI patients have appropriate dynamic flexion/extension cervical radiographs performed, thus underestimating the association of VBI/PCS with CVJ anomalies.\(^6\)

This case was first presented with PCS at the age of 8 years and underlying cause was missed in initial neuroimaging as attendants refused for detailed MRI evaluation with angiogram. It was later diagnosed when PCS recurred at the age of 13 years, presented as locked-in syndrome due to pontine infarct. After extensive review of literature, we found that BI causing stroke in children is extremely rare in comparison to some cases reported in adults, but of great clinical significance.\(^6\) Because natural infarct evolution is to be expected, repeat neuroimaging may be necessary in such cases.\(^7\)

Diagnostic tools for BI are lateral x-ray with odontoid views, CT scan, MRI and somatosensory evoked potentials.\(^3\)\(^8\) In ischemic stroke, diffusion weighted imaging have reported sensitivity of 90-98% depending on the stroke location and timing of the study. CT and MR angiography are comparable in identifying vascular involvement in PCS, but have limitations. Conventional catheter cerebral angiogram remains the gold standard.\(^7\)

Currently, no defined treatment guidelines exist for BI and surgical treatment continues to centre on varying combinations of anterior (endonasal and transoral) or posterior (occipital-cervical) decompression with or without traction or fusion, with high rate of mortality and morbidity.\(^3\)\(^4\)

**CONCLUSION**

In conclusion, all young patients presenting with features of VBI or PCS should be screened for CVJ anomalies, so that early interventions can be carried out without further neurological damage.

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**REFERENCES**
