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OCULOMOTOR PALSY IN A CASE OF SCHIZENCEPHALY: A RARE CASE REPORT

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ABSTRACT

Schizencephaly is a rare congenital disorder of brain development characterised by cerebral cerebrospinal fluid(C.S.F) containing clefts connecting the ventricles to the subarachnoid space with a reported incidence of 1.48 in 100,000 population. The usual presentation is hemiparesis, epilepsy, mental retardation or septo-optic dysplasias. Till date, cranial nerve palsy has never been ascribed to schizencephaly. Here we report a case of oculomotor nerve palsy in a case of schizencephaly.

INTRODUCTION:

Schizencephaly-a term, first used by Yakovlev and Wadsworth in 1946(1) refers to a congenital disorder of cerebral cortical organisation characterised by the presence of deep clefts connecting the ventricular ependyma with the Pia mater.(2)

Its prevalence is estimated to be 1.48 to 1.54 in 100,000 population and 1 in 1650 children with epilepsy(3).

There is no known sex predilection(4).

It is basically of 2 types:

- Type 1(closed lip): with narrow clefts (pia-ependymal seam) and juxtaposed borders at some places, which is less common(5).
- Type 2(open lip): with wide clefts encompassing a significant amount of CSF, which is more common(5).

Macroscopically, the clefts in Schizencephaly can be unilateral or bilateral, mostly involving the fronto-parietal area and/or perisylvian region.(5)

Microscopically the clefts are lined by heterotopic, polymicrogyric grey matter.

The cause of disorganised development of grey matter is not fully understood. Reported aetiologies include cytomegalovirus(6, 7),several types of maternal trauma(8) and a myriad of other possible etiologies have been propounded such as, toxic, metabolic, vascular(9) and other infectious insults caused during the third and fourth month of antenatal period of neuroblast migration(10).

Familial cases have also been reported(11).

Genetically, latest reports suggest that EMX2 gene (located on 10q26.1 chromosome) mutations are not a common cause of schizencephaly(12), contrary to the earlier presumptions(7).

Maternal ultrasonography can be used to diagnose the condition antenatally.

MRI is more sensitive than Computed Tomography(CT) in detecting the clefts as well as accompanying abnormalities such as hydrocephalus, septo-optic dysplasia, cysts, dandy-walker malformation(13).

Treatment is directed mainly towards the control of seizures either by medications or by surgical resection of epileptogenic focus(14) and physiotherapy for the paretic limb.

Case report:

Here is a 40 years old, male, who presented to our outpatient department in a drowsy state with a history of low grade, intermittent fever and progressively deteriorating consciousness for last 8 days. He had an antecedent episode of partial seizure with secondary generalisation. History of headache and occasional episodes of vomiting was also present.There was no history of head trauma, any drug intake, any substance abuse or any ongoing systemic illness. His caretakers gave past history of mild mental retardation, right sided body weakness since birth and seizure disorder and right eyelid drooping since childhood for which he was never investigated much. The seizure episodes used to be of generalised tonic clonic nature, very brief in duration and with postictal confusion lasting 5-10 mins without any loss of consciousness.The family history was insignificant.

After giving 3 days of anti-epileptic treatment, he regained consciousness. Upon examination we found moderate intellectual disability, ptosis of right eyelid with absent direct and consensual pupillary reflexes and all the eye movements were restricted except abduction and downward gaze. Tone was increased in right half of body along with slightly reduced power, brisk deep tendon reflexes and a positive babinski's sign. Rest of the examination was unremarkable.

Results of all routine biochemical investigations were within normal limits.

The HIV and Malarial parasite test were also negative.

Then we proceeded on with a lumbar puncture. The Cerebrospinal fluid(C.S.F) findings are noted in table 2 (table 2).

Then MRI of brain was done (figure 1,3).

DISCUSSION:

Schizencephaly is a congenital disorder, therefore the motor and intellectual disabilities are usually present since birth but in some cases late presentation of symptoms especially seizures has also been reported. The extent of motor and mental disabilities are reported to be directly related to the extent of malformation.(15)

Our case had a large cleft on the left side(figure 1)

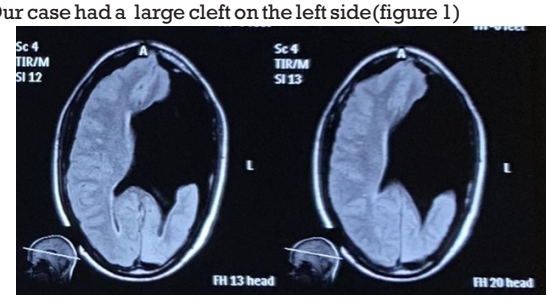

Figure 1: T2W_FLAIR image showing left sided large schizencephalic cleft.

The cleft was of open lip variety associated with an arachnoid cyst in posterior cranial fossa. Moderate degree of mental retardation and right sided spastic hemiparesis was present since birth, which was congruent with the size of the cleft.

Our patient also had right sided ptosis(figure 2) associated with absent pupillary light and accommodation reflexes along with restricted adduction, downward and upward gaze in the right eye.

Figure 2: Ptosis in right eye.

The lateral gaze was normal. All these findings suggested a weakness of oculomotor nerve in the right eye. However, the history of onset of both ptosis and epilepsy pointed back to childhood and not from birth. The cause of this late onset oculomotor weakness may be compression of midbrain by the enlarged right temporal lobe which is evident in the M.R.I. The enlarged temporal lobe may be a part of the disorganised brain development.

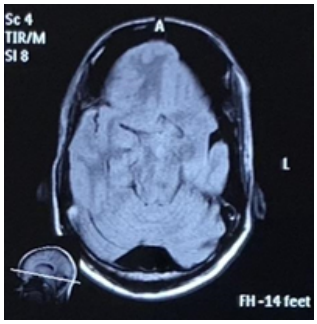

Figure 3: T2W-FLAIR image showing relatively large right temporal lobe.

The other differential for oculomotor weakness could be, as a sequelae of an encephalitic episode which may have occurred in childhood but there was no clear history suggestive of such an episode.

The present episode of longer than usual alteration in consciousness following seizure episode seemed to be due to associated meningo-encephalitis, which was supported by the history of fever coupled with raised CSF protein and low sugar, though without pleocytosis (table 1).

Table 1: Cerebrospinal fluid(C.S.F) values at presentation.

CSF	Value
Appearence	Clear
Total cell count(/mm^3)	4
Polymorphs	0
Lymphocytes	4
Red blood cells	0
Protein(mg/dl)	115
Sugar(mg/dl)	49
Adenosine deaminase(u/l)	3.7

Gram's staining and culture of C.S.F was negative.Further investigations for finding the etiological agent was not possible due to lack of resources.

The patient was followed up after 6 weeks with a repeat lumbar puncture which showed normal C.S.F values but the oculomotor palsy still persisted to the same level.

We found no such case of schizencephaly having associated cranial nerve palsies in our literature search. So this presentation makes it an important case worth notifying.

Conclusion: Thus we conclude that cranial nerve palsy can also be a manifestation of schizencephaly along with the other more commonly reported features.

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