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Case Report

Radiology Section

Hydranencephaly in a Child with Congenital Cytomegalovirus Infection- A Clinicoradiological Perspective

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ABSTRACT

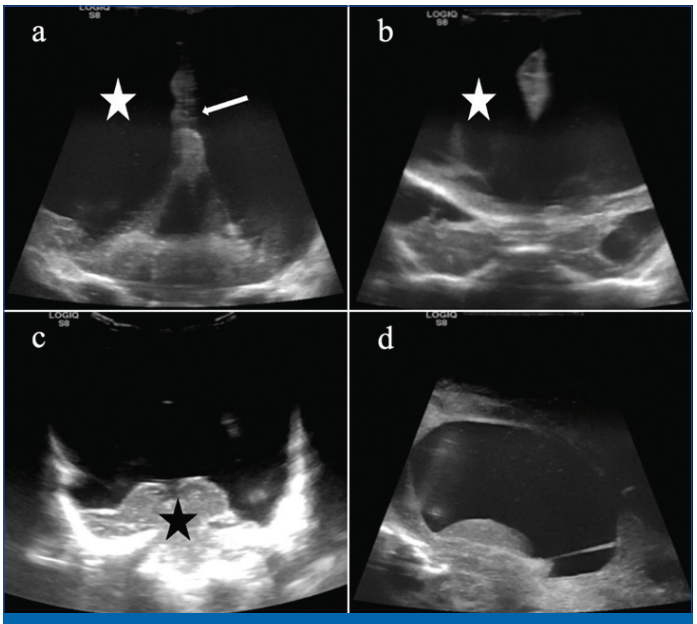
Hydranencephaly (HE) is a rare congenital anomaly in which there is a complete or near complete absence of bilateral cerebral hemispheres which are replaced by a membranous sac filled with Cerebrospinal Fluid (CSF). Thalami, brainstem, and cerebellar hemisphere are preserved. A small island of cerebral parenchyma may also be seen along the inner aspect of the calvarium. Several theories regarding its aetiopathogenesis have been postulated with the hypothesis of bilateral occlusion of supraclinoid segment of Internal Carotid Artery (ICA) being widely accepted. The authors present a case of a two-month-old male child presenting with increasing head size since birth, associated hydrocephalus and residual parenchymal sleeve visualised on neurosonogram and calcifications depicted on computed tomography. The diagnosis of Cytomegalovirus (CMV) was confirmed serologically and further neurosurgical consultation was advised for increasing head size secondary to hydrocephalus. The HE is usually detected in the second trimester, however can also occur in early or later gestation and has a very poor prognosis.

Keywords: Calcification, Falx cerebri, Neurosonogram

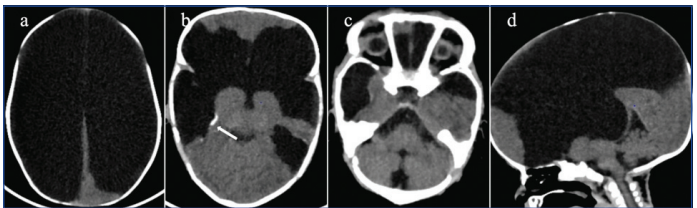
CASE REPORT

A two-month-old male child presented to the Paediatrics clinic with complaints of a gradual increase in head size as observed by parents since birth. In addition, the child had poor suckling reflex and persistent intermittent vomiting. The child was born out of a non consanguineous marriage and born at term by normal vaginal delivery. There was no history of any symptoms related to infection in the mother while she was bearing the child. The mother neither attended any antenatal check up nor undergo an ultrasound scan during the entire antenatal period.

On examination the child was conscious, crying with the presence of sunset sign. Anterior fontanelle was patent and bulging. Anthropometry examination revealed a weight of 4 kg, a height of 52 cm, and a head circumference of 40 cm (>97 percentile for age). A neurosonogram was requested which showed near-complete replacement of cerebral parenchyma by CSF with preservation of some parts of frontal, temporal, and occipital lobes, intact falx cerebri and posterior fossa structures, and [Table/Fig-1]. Non Contrast Computed Tomography (NCCT) of the head confirmed enlarged head size along with the presence of calcification on the surface of residual brain parenchyma raising suspicion of intrauterine Cytomegalovirus (CMV) infection [Table/Fig-2]. Among laboratory findings, Anti CMV Immunoglobulin M (IgM) serological test was sent which was positive. The normal course of pregnancy related to mother during pregnancy along with positive CMV serology in newborn child suggest asymptomatic manifestation of the disease. Magnetic Resonance Imaging (MRI) brain confirmed neurosonogram findings of residual cerebral parenchyma, intact falx cerebri, and preserved posterior fossa structures [Table/Fig-3]. Since, he was the firstborn child, a history of similar disease in the sibling was not applicable in our case. The parents were counselled regarding the poor outcome of the condition and urgent neurosurgical referral was suggested. Neurosurgery consultation was done and as residual cortical mantle was less than 1 mm in thickness no active intervention was suggested. The prognosis and expected future complications including aspiration were explained to the caregivers. At the time of discharge, the patient was conscious, accepting feeds and moving all limbs.



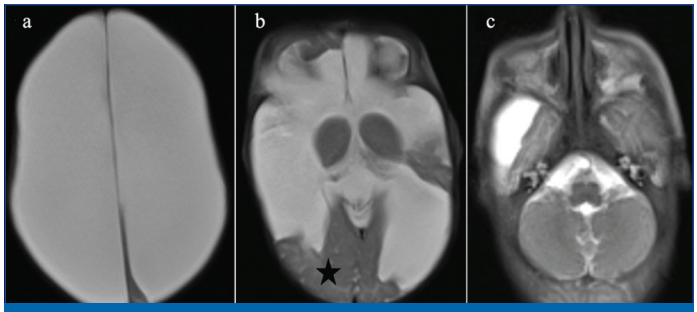
[Table/Fig-1]: Trans fontanelle coronal (a-c) and midsagittal (d) neurosonogram images show replacement of cerebral parenchyma by CSF (white asterisk in a,b) with preserved falx (white arrow in a) and posterior fossa structures (black asterisk in c).



[Table/Fig-2]: Non contrast CT scan axial (a-c) and sagittal (d) sections of the brain shows near-complete replacement of cerebral parenchyma with CSF, the preserved island of bilateral frontal, temporal and occipital lobe parenchyma. Focal calcification is seen along the surface of the right thalamus (white arrow in b).

DISCUSSION

Hydranencephaly is the severe form of cerebral cortical anomaly affecting less than 1 per 10,000 live births [1]. It is characterised by the complete or near complete absence of cerebral hemispheres which are replaced by a membranous sac filled with CSF, glial



[Table/Fig-3]: T2 weighted MRI axial images confirm the CT findings with preserved occipital brain parenchyma (black asterisk in b), brain stem and cerebellum.

tissue, and ependyma with preservation of the overlying skull [2]. Prognosis is very poor leading to intrauterine foetal demise in the majority. Rarely, it is seen in postnatal life, with death occurring in first year of life. There is no significant risk of this entity recurrence in subsequent pregnancies [3].

The HE is a rare neurological disorder characterised by the absence and replacement of the cerebral hemisphere by a large CSF pool. Irrespective of underlying aetiology the cerebral brain parenchyma undergoes liquefactive necrosis and eventual replacement by CSF, however, the overlying meningeal covering remains intact [4]. The exact cause is not known; however, the following aetiologies have been described:

Infarction: Bilateral occlusion of the supraclinoid segment of the internal carotid arteries or middle cerebral arteries, is the most accepted hypothesis [5].

Infection: Necrotising vasculitis or local destruction of the brain tissue secondary to intrauterine infections like CMV, toxoplasmosis, rubella, and Herpes Simplex Virus (HSV) [6-8].

Thromboplastic material from a deceased co-twin: Monochorionic twins have presented with a variety of cerebral lesions in the recipient twin resulting from emboli or thromboplastic material originating from the macerated co-twin [9].

Diffuse hypoxic-ischaemic brain necrosis: Foetal hypoxia secondary to maternal exposure to carbon monoxide or butane gas may result in massive tissue necrosis with cavitation and eventual resorption of necrotised tissue [2].

Apart from the above group of implications, specific maternal factors including young age at the time of pregnancy and substance abuse are also known causative factors.

In intrauterine life, foetal movements are perceived within normal limits with unremarkable course except for the detection of imaging changes detected during antenatal ultrasonography [2]. At birth, a child with HE may seem unremarkable or show the skull and or facial asymmetry. Spontaneous reflexes may seem intact at first, progressing to muscle rigidity and irritability. Later in the course of the disease, the child may present with seizures, visual or auditory symptoms, growth disturbance and motor abnormality of limbs [4]. The HE commonly affects both cerebral hemispheres, with rare forms affecting only one cerebral hemisphere inducting a better prognosis [10].

Neurosonogram, NCCT and MRI are commonly used to reach the diagnosis. The HE is commonly diagnosed between the 13th and 26th weeks of gestation, as the concerned structures are discernible by this period of gestation [2]. Neurosonogram commonly shows near-complete replacement of enlarged cranial cavity with fluid contents, absent or barely present cerebral parenchyma and absence of midline falx. Regarding the pattern of calcification, intrauterine infections commonly occur in periventricular locations, albeit in our case due to non visualisation of the ventricular system the same could not be appreciated.

An MRI with Magnetic Resonance Angiography (MRA) is the best imaging modality overall as it aids in the detection of cortical remnant and also evaluates for ICA stenosis, explaining the commonly accepted hypothesis as well as allowing definite exclusion of other differentials.

The prognosis is very poor with the majority of cases showing foetal demise in-utero and the rest of children passing away within the first year of life. Among those who survive, suffer from refractory seizures, limb spasticity, respiratory infections and growth retardation [1,3]. Prolonged survival is very rare [3]. The treatment is supportive and aimed at the treatment of any aforesaid complications with the main emphasis on the management of hydrocephalus by placing a shunt and preventing complications related to long term shunt placement [2,4]. Supportive care is also required for the neurological and respiratory systems in the long run.

Differential diagnoses can include cases of severe hydrocephalus, porencephalic cyst and alobar holoprosencephaly. In severe hydrocephalus, a thin cortical rim of the brain parenchyma is always seen. In alobar holoprosencephaly, there are usually coexisting midline facial malformations and smaller size of the head, with the absence of falx cerebri. Porencephalic cysts are usually located in the middle cerebral artery territory secondary to ischaemic infarcts resulting in localized areas of cortical gliosis and showing a similar presence of cerebral mantle as above two conditions [11]. An MRI can aid to define imaging manifestations in cases with a diagnostic dilemma as well as serve as a confirmatory modality [4].

As far as associations are concerned intracranial calcifications, cerebellar and choroidal hypo/dysplasia and migrational anomalies were encountered in the past. Beyond neural abnormalities, other organ systems involvement is also seen like genitourinary and musculoskeletal systems [2,12]. Recently introduced white matter abnormalities in the continuum of congenital CMV infections include cleft cortical dysplasia and schizencephaly [13]. In our case, the diagnosis of HE secondary to congenital CMV infection was made based on imaging findings of associated intracranial calcification supported by a positive serological test.

CONCLUSION(S)

Hydranencephaly is a rare disease leading to in-utero death and a very poor life expectancy. The importance lies in early detection by antenatal imaging, terminating pregnancy at earliest possible gestation. Timely diagnosis and obviating the continuation of gestation helps in reducing maternal morbidity and mental disharmony, albeit with some ethical concerns. Those surviving beyond one year of life will either die in early infancy or maybe persistent in a vegetative state beyond this period. In established cases, counselling of prospective parents is required for timely antenatal imaging and serology for potential virological agents reducing the occurrence in future gestations.

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