Letter to Editor

Hydranencephaly: A Rare Cause of an Enlarging Head Size in an Infant

Dear Editor,

Hydranencephaly is a rare congenital abnormality characterized by absence and replacement of the cerebral hemispheres by a large cerebrospinal fluid pool. It is thought to be caused by occlusion of bilateral internal carotid arteries in the fetal life mainly during the second trimester due to a variety of causes.[1,2] It is one of the recognized forms of brain malformations which is usually associated with intrauterine fetal demise and is therefore rarely seen in postnatal life.[3] It is a rare entity with a reported incidence of less than 1 per 10,000 live births.[4]

A 4-month-old preterm infant was brought by his parents to the pediatric outpatient department with complaints of recent onset of fever and progressively enlarging head size. The fever was of moderate degree and was not associated with chills and rigors. According to the parents, the size of the baby's head was normal at the time of birth but it started increasing after one week of life. On examination, the child was lethargic with increased occipito-frontal circumference and there was hypotonia in all the four limbs. There was no associated cranio-facial dysmorphism or limb deformity. The mother also reported that the eyes of the child showed roving movement. The primitive reflexes, such as grasp and suction, were intact. He responded only to sound and painful stimuli and a lack of awareness to the surroundings was noted. The fundus examination of the child was normal. The mother was neither diabetic nor hypertensive; she was not a smoker and had never ingested alcohol. There was no history of radiation exposure or febrile illness and exanthematous skin rash during pregnancy. She did not undergo any ultrasound examination during the antenatal period. There was no history of consanguinity or similar complaints in the other siblings. After admission, he was managed with a working diagnosis of complicated tubercular meningitis. Mantoux test and CSF examination were normal. The baby was then referred for computed tomography (CT) of the head.

Axial CT of the head showed complete absence of B/L cerebral hemispheres with no cortical mantle and their replacement by fluid [Figure 1]. However, entire falx was preserved excluding holoprosencephaly. Also thalamus, pons, medulla, and cerebellar hemispheres were relatively preserved [Figure 2], all in keeping with the diagnosis of hydranencephaly. Somatosensory evoked potentials (SSEP) showed complete absence of the cortical activity with preservation of waves of brainstem origin. Electroencephalogram revealed a flat pattern. The visual evoked potential was absent bilaterally. The parents of the baby were counseled for poor prognosis of the condition and the minimal risk of recurrence in subsequent pregnancies. They refused any active surgical intervention.

Hydranencephaly is a rare encephalopathy that occurs in-utero and presents rarely in postnatal period. The posterior fossa structures as well as parts of the temporal and occipital lobe supplied by posterior cerebral arteries are usually preserved.[1,5] However, cases with cerebellar hypoplasia and occlusion of bilateral posterior cerebral arteries have been reported.[1] This rare disorder usually occurs after ventricles have already formed; hence, the falx is preserved. Hydranencephaly is associated with various congenital anomalies, including arthrogryposis, renal aplastic dysplasia, poly-valvular heart defect, trisomy 13 and Fowler syndrome.[6]

Most common etiologies described for hydranencephaly are occlusion of the supra-clinoid segment of bilateral internal carotid arteries,[1] or an extreme form of leukomalacia formed by confluence of multiple cystic cavities. Other etiologies include fetal hypoxia due to maternal exposure to carbon monoxide or butane gas resulting in massive tissue necrosis with cavitation and resorption of necrotized tissue and necrotizing vasculitis or local destruction of the brain tissue secondary to intrauterine infections, e.g. congenital toxoplasmosis, cytomegalovirus and herpes simplex.[4,7]

Hydranencephaly can be diagnosed using ultrasound (ante- and postnatal), Magnetic Resonance Imaging (ante- and postnatal) and CT. The radiological features include almost complete absence of cortical mantle. The cranium is completely filled with membranous sac containing cerebrospinal fluid. The skull may be normal-sized or enlarged as was seen in the present case. The falx cerebri and tentorium cerebelli are usually intact. Thalamus, hypothalamus and choroid plexus are preserved and cerebellum is

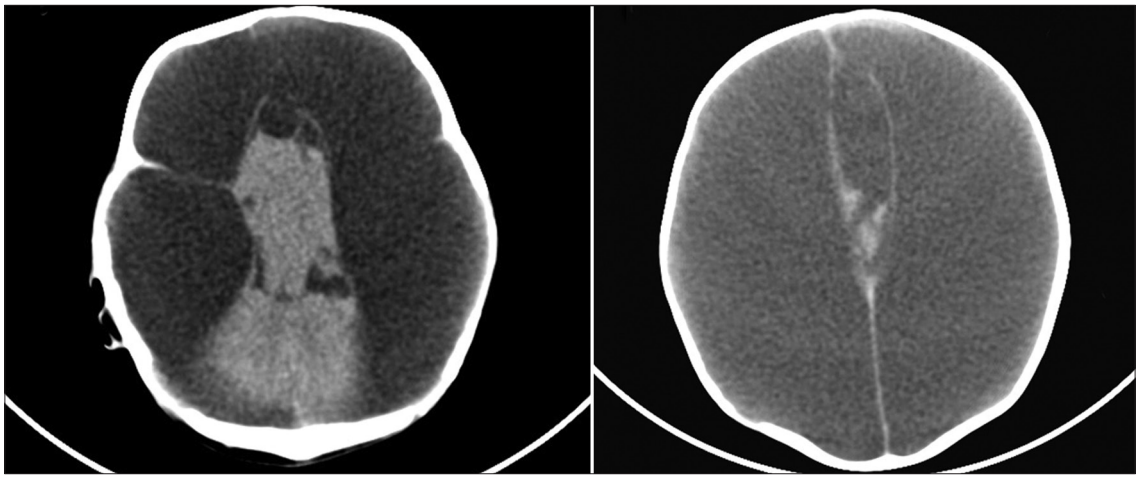

Figure 1: Non-contrast axial CT image shows (a) complete destruction of bilateral cerebral hemispheres with no evidence of cortical mantle with preserved midbrain and (b)falx cerebri

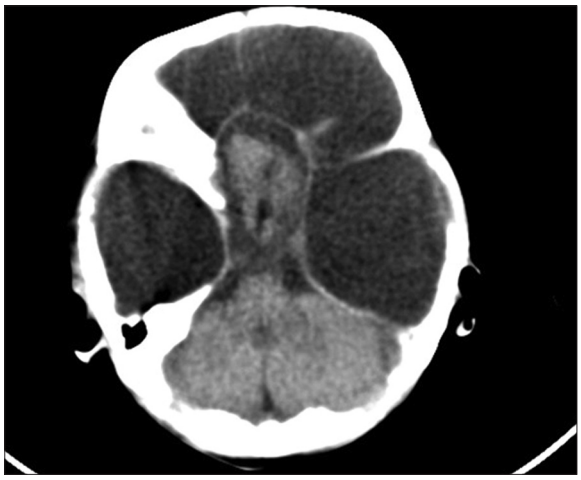

Figure 2: Non-contrast axial CT image shows relatively normal cerebellar hemispheres, brainstem, and bilateral thalami

intact.[1,4] The electrophysiological studies confirm the imaging findings. Prolonged survival upto 22 years has been reported in literature with significant associated morbidity.[3,8] Ventriculo-peritoneal shunting has been tried with limited success because of frequent need for shunt revision or removal and associated poor prognosis for psychomotor development.[9] Recently, endoscopic choroid plexus coagulation is being explored as an alternative to shunting.[10]

The condition needs to be differentiated from severe obstructive hydrocephalus and severe open lip schizencephaly, however, in both these conditions, the cortical mantle is only reduced in thickness. Alobar holoprosencephaly can be differentiated by the presence of falx and frequent coexisting midline facial abnormalities.

Thus, hydranencephaly should also be considered in the differential diagnosis of an infant presenting with enlarging head size, especially in children with psychomotor retardation.

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