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INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI: 10.21474/IJAR01/12497
DOI URL: http://dx.doi.org/10.21474/IJAR01/12497

ISSN NO. 2320-5407

RESEARCH ARTICLE

HYDRANCEPHALY : RARE ENTITY WITH INCIDENCE LESS THAN 1 IN 10000 LIVE BIRTHS

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Manuscript Info

Manuscript History
Received: 15 December 2020
Final Accepted: 18 January 2021
Published: February 2021

Key words:-
Hydranencephaly, Enlarging Head Size, Absent Cerebral Cortex, Magnetic Resonance Imaging (MRI), Ultrasonography(USG), Computed Tomography (CT)

Abstract

Hydranencephaly is a rare entity with incidence of 1 in 10000 live births and is characterized by near total absence of cerebral cortex and basal ganglia which are replaced by cerebrospinal fluid. Various aetiologies of hydranencephaly have been suggested which include: Infarction, leukomalacia, diffuse hypoxic-ischemic brain necrosis, intrauterine infections leading to necrotizing vasculitis. Ultrasonography, Computed Tomography and Magnetic Resonance Imaging can easily detect and diagnose hydranencephaly. We present a case of 2-month-old term infant diagnosed with hydranencephaly on computed tomography.

Introduction:-

Hydranencephaly is characterized by near total absence of cerebral cortex and basal ganglia which are replaced by cerebrospinal fluid. Thalami, pons ,cerebral peduncles, cerebellum are usually present and rarely small amount of tissue from occipital, frontal, temporal lobes is also present. Its incidence is less than 1 per 10,000 live births[1]. It is a rare congenital abnormality with very less cases seen in postnatal life[2]. Most common etiology of hydranencephaly is occlusion of bilateral internal carotid arteries in the second trimester of pregnancy leading to intrauterine fetal demise[3,4].

Case:

A 2-month-old term infant with complaints of recent onset progressively enlarging head size came to hospital for paediatric consultation .The size of the baby's head was normal at the time of birth but it started increasing after 2nd week of life .When baby was examined by the paediatrician, the head circumference was greater than normal with bulging anterior fontanelle, sluggish sensory motor reflexes, hypotonic limbs, weak cry. During the intrauterine life, there was no history of maternal diabetes, hypertension, fever, skin rash, smoking, radiation exposure, alcohol, teratogenic drug intake. Provisional diagnosis of hydrocephalus was made and baby was referred to radiology department for computed tomography (CT) of the head.

Axial CT of the head showed absence of bilateral cerebral hemispheres with no cortical mantle and their replacement by fluid , only small amount of cerebral tissue in temporal and occipital region was present [Figure 1a,1b]. Thalamus, pons, medulla and cerebellar hemispheres were relatively preserved [Figure 2]. Falx was present which differentiate the fluid filled supratentonial space of hydranencephaly from holoprosencephaly. Based on above findings the diagnosis of hydranencephaly was made.

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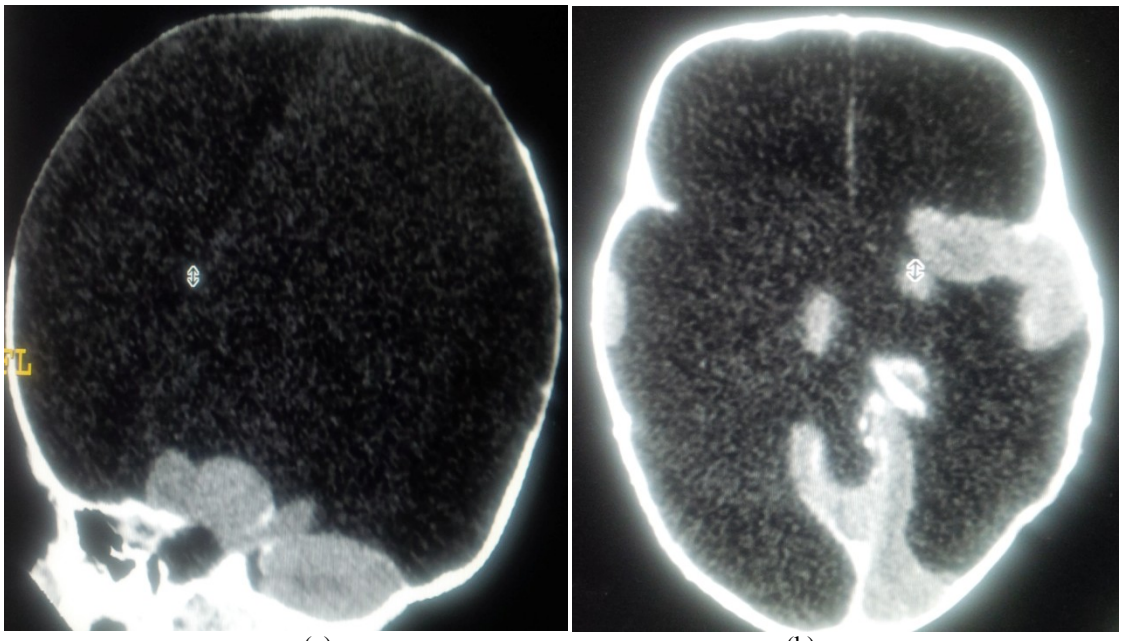

(a) (b)

Figure 1:- a. Sagittal CT scan image shows absent cerebral cortex which is replaced by CSF. Thalamus, hypothalamus and cerebellum are preserved. Figure 1b. Axial CT image showing falx cerebri, remnant of temporal and occipital lobes.

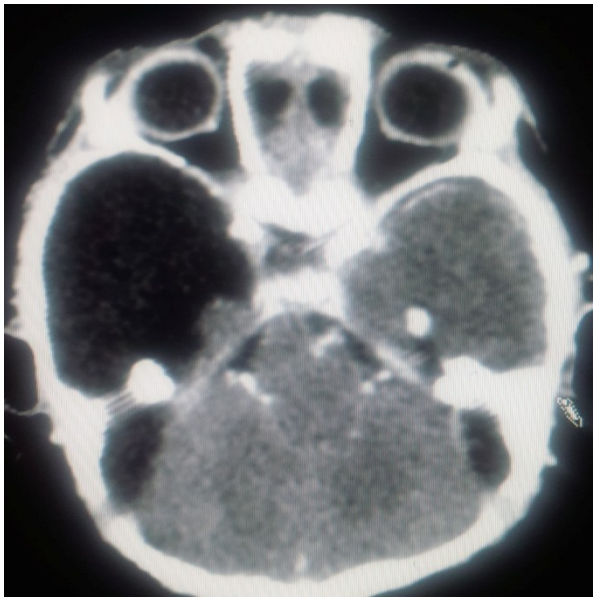

Figure 2:- Axial CT image showing preserved cerebellum, brain stem structures and part of temporal lobe.

Discussion:-

The exact cause of hydranencephaly is not clear. However, various aetiologies have been suggested which include: Infarction due to occlusion of the supraclinoid segments of the internal carotid arteries or of the middle cerebral arteries, leukomalacia, diffuse hypoxic-ischemic brain necrosis from fetal hypoxia due to maternal exposure to carbon monoxide or butane gas resulting in massive tissue necrosis with cavitations and resorption of necrotized tissue, intrauterine infection leading to necrotizing vasculitis. Intrauterine infections associated with hydranencephaly include congenital toxoplasmosis, cytomegalovirus, and herpes simplex infections[5].

Congenital anomalies like arthrogryposis, renal aplastic dysplasia, poly-valvular heart defect, trisomy 13 and Fowler syndrome are associated with hydranencephaly[6].

During the intrauterine fetal life imaging modalities such as Ultrasonography, MRI and Computed tomography (CT) during postnatal life can easily detect and diagnose hydranencephaly . The radiological features are almost same in all these imaging modalities and include almost complete absence of cerebral cortex which is replaced by membranous sac containing cerebrospinal fluid. The cortical mantle is also absent. The falx cerebri and tentorium cerebelli are preserved. Thalamus, hypothalamus, choroid plexus and cerebellum are preserved[1,4].

Differential diagnosis for this condition include severe obstructive hydrocephalus and open lip schizencephaly, however, in both these conditions, the cortical mantle is only reduced in thickness. In alobar holoprosencephaly falx cerebri is absent.

Thus, hydranencephaly is an important differential diagnosis in an infant presenting with enlarging head size and psychomotor retardation and should be evaluated radiologically for early establishment of definitive diagnosis and further management.

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