ORIGINAL RESEARCH PAPER
Volume - 10 | Issue - 04 | April - 2021 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

HYDRANENCEPHALY – RARE CASE REPORT AND ITS RADIOGRAPHIC FEATURES

Radiodiagnosis

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ABSTRACT

Hydranencephaly is a congenital disorder characterized by the replacement of the cerebral parenchyma with a CSF filled membranous sac. Posterior fossa structures are usually not involved. Various etiologies have been suggested, among these ischemic strokes due to bilateral ICA or MCA occlusion is more common. At birth, clinical findings would be large sized head, normal brainstem reflexes and cortical activity. In the following weeks, infant may show increase in muscle tone, become irritable and present with seizures. Other symptoms include blurring /loss of vision, loss of hearing, growth retardation, weakness in the limbs and intellectual deficits. Neonate was referred to the department of radiology on postnatal day 3 for imaging evaluation of a case with a clinical suspicion of Hydranencephaly. Computed tomography was done to confirm the diagnosis. Hydranencephaly was confirmed after radiological evaluation. Treatment is usually palliative. Early antenatal detection helps in the termination of pregnancy. Prognosis is poor in these patients and most have the life span of about 12 months.

KEYWORDS

Antenatal; Cerebral parenchyma; CSF filled membranous sac; Hydranencephaly; Neonate.

INTRODUCTION:

Hydranencephaly, a rare congenital disorder characterized by replacement of cerebral parenchyma with a CSF filled membranous sac. Owing to earlier normal embryogenesis of the telencephalon, the meninges and the skull are well formed. The cranial cavity may contain glial and ependymal tissues, more commonly close to diencephalon and along the falx. There is relative sparing of the posterior fossa structures. Incidence rate is 1 per 10,000 births.

Various etiologies have been postulated like bilateral ICA or MCA occlusion, in-utero infections by toxoplasmosis and few viruses. Bilateral MCA and ICA occlusion are the most common.

Patients at birth may show normal growth and primitive refluxes. Later in the life, they present with large sized head due to increased intracranial CSF pressure. Diagnosis can be made during antenatal ultrasound. The prognosis is poor in children with hydranencephaly. Most patients with this anomaly die before 1 year of birth. However, in some, they survive for several years.

Treatment is palliative. Patients with the hydrocephalus are considered for ventriculo-peritoneal shunt.

Case Presentation:

A 24 years old female, Gravida 1 Para 0 Abortion 0, visited our hospital at 34 weeks +5 days of gestational age with bleeding per vagina and abdominal pain. She was registered in a district hospital, received antenatal care as per routine, anomaly scan was done at 18 weeks which revealed no gross congenital anomalies. Her first and second trimester gestations were uneventful.

Past History:

Non-diabetic and normotensive. She had taken routine antenatal drugs like Iron and folic acid tablets and had not been exposed to teratogenic or radioactive agents.

Family History:

No significant family history of genetic disorders

Clinical Assessment:

Vitals: Pulse – 84 bpm, Blood pressure – 100/70 mm Hg; Respiratory rate – 20/min; Temperature – 99.2 F.

Abdominal Examination:

Uterus was of 36 weeks size; in longitudinal lie and Cephalic presentation, Fetal Heart Rate (FHR) was recorded to be 140 bpm, Contractions - one in ten minutes.

Screening Antenatal Ultrasound:

i) A single intrauterine live fetus of 35+2 weeks, ii) Anterior placentation, Grade III maturity, iii) Liquor: adequate, iv) Fetal head showed a large intracerebral cavity filled with anechoic fluid. Mildbrain and cerebellum appeared normal. With the above features, a diagnosis of hydranencephaly was suggested.

Fetus was delivered by Emergency Caesarean section as a result of cephalopelvic disproportion. For 2 minutes following birth, Baby did not cry. Nasal and oral suctioning/tactile stimulation/ BMV were performed. The 1 min APGAR score was 2/10 and 5 min score was 8/10. Birth weight recorded was 3.0 kg and Head circumference at birth was 39 cm (> 97th percentile for age).

Neonate was then shifted to NICU for further management of respiratory distress. Baby was on Nasal CPAP for 2 days and was weaned from it on 3rd day.

Head Examination:

Large sized trans-illuminant head along with widely open fontanelles.

Systemic Examination: Intact reflexes.

Neonatal investigations performed included Neurosonogram, USG abdomen and Echocardiogram. Postnatal USG Cranium findings were consistent with screening antenatal ultrasound done at the time of admission.

Cardiac and Renal anomalies were not found in this patient.

Laboratory evaluation for sepsis revealed negative results.

Mother had been investigated for TORCH group agents infection and

results came as negative.

Parents were counseled on the diagnosis and the prognosis.

On postnatal day 3, patient was referred to the Department of Radio-diagnosis to take Radiograph of Head (Figure1) and Computed Tomography of Brain – Bone window, Brain window and (3D) VR (Figure 2-5) for confirmation of the diagnosis.

Neonatal Head Radiographs Revealed:

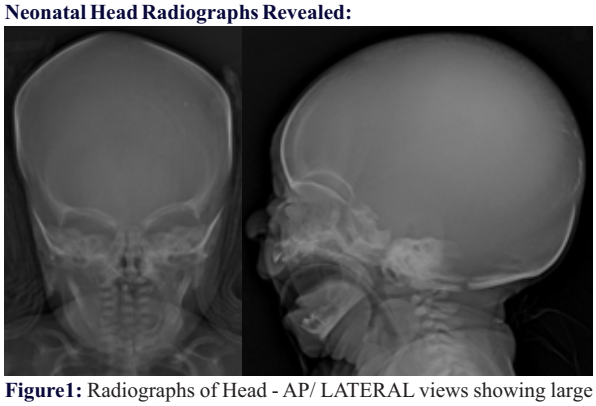

Figure1: Radiographs of Head - AP/ LATERAL views showing large sized head with thin skull bones, widened fontanelles and J shaped sella, while the facial bones appear normal.

Computed Tomography Revealed:

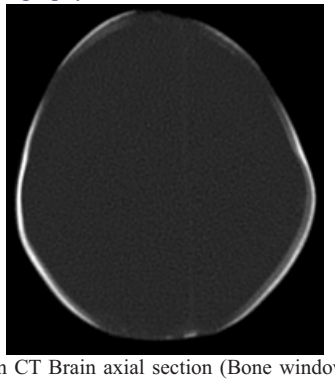

Figure 2: Plain CT Brain axial section (Bone window) - Increased head circumference with severe thinning of both outer and inner tables of the calvarium and wide fontanelles.

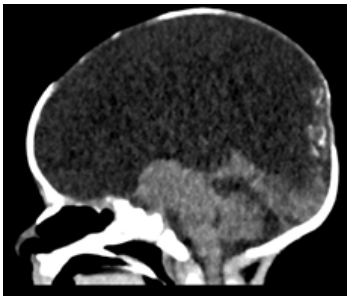

Figure 3: Plain CT Brain Mid-Saggital section (Brain window) – CSF density near completely replacing the cerebral cortex with remnant cerebral cortex and posterior falcine hyperdensities.

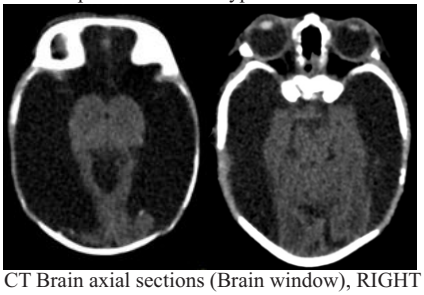

Figure 4: CT Brain axial sections (Brain window), RIGHT - Normal bilateral basal ganglia, thalami, LEFT - Normal Midbrain and cerebellum with remnant adjacent cortices.

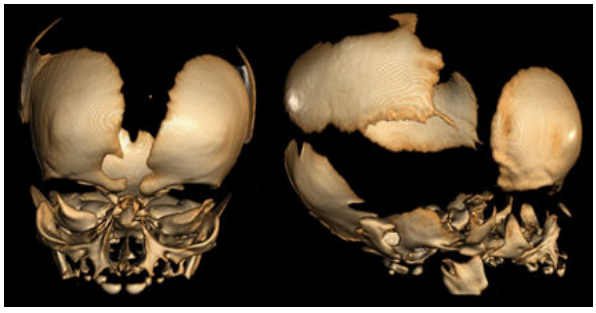

Figure 5:3D Virtual Reconstruction (VR) Images – Increased head circumference with severe thinning of the calvarium and widened fontanelles.

Patient was managed conservatively and given palliative treatment with Vitamin D3 supplementation 1ml OD upto 6months of age, warmth, exclusive breast feeding upto 6 months of age and regular immunization. Patient was given counseling and advised for regular follow-up in Neurology department. Patient understood the condition of the baby, prognosis of the child and is willing to adhere to the palliative treatment & regular follow-up.

DISCUSSION:

Hydranencephaly is a rare post-neurulation congenital disorder occuring during the 2nd trimester. This disorder occurs following neural migration and prior to synaptogenesis. There is destruction of the normal cerebral parenchyma developed in-utero owing to the encepahaloclastic process.

It is one of the most severe forms of bi-hemispheric cortical anomalies in which the cerebral hemispheres are show near total replacement by a CSF filled membranous sac, ependyma and glial tissue. The midbrain, cerebellum, thalami, basal ganglia, choroid plexus and skull bones are not usually involved. Sometimes only one cerebral hemisphere might be involved which is known as Hemihydranencephaly (HHE).

Various etiologies have been postulated –
a)Occlusion of bilateral ICA or MCA leading to infarction of structures perfused by the ICA/MCA,
b)In-utero TORCH infections,
c)Fetal hypoxia may cause diffuse hypoxic-ischemic brain necrosis,
d)Maternal toxin exposure which include such as smoking, cocaine, sodium valproateand estrogens,
e)Fowler syndrome – A rare genetic disorder characterized by proliferative vasculopathy, hydranencephaly-hydrocephalus complex) and deficiency of factor XIII.

Most of the cases are diagnosed during regular 2nd trimester antenatal ultrasound. Follow-up can be advised to confirm the diagnosis. If this disorder is not detected antenatally, diagnosis is delayed as the neonate may appear normal. Gold standard modality to diagnose this condition is MRI of the Brain. This helps in differentiating this from Severe Hydrocephalus and Holoprosencephaly.

Various modalities used in the diagnosis are –
1. Ultrasound: Anomaly ultrasonography done during weeks 19 to 23 of gestation reveal absence/ non visualization of bilateral cerebral hemispheres, which are replaced by anechogenic fluid collection with homogenous echogenic material along the falx and posterior fossa. There is relative preservation of cerebellum, brainstem and thalami. Post natal ultrasound is also used to diagnose this condition.

2. Brain MRI: This is the gold standard in diagnosing this condition. It shows the absence/ non visualization of supratentorial cerebral parenchyma, near totally replaced by a CSF filled membranous sac. Remnants of parenchyma, predominantly along the occipital lobe, may be noted. Falx, basal ganglia, brainstem and cerebellar hemisphere are normal.

3. Brain Computed tomography: This shows similar findings as seen in Brain MRI.

4. Other ancillary tests performed are:
- Loss of electrical activity on Electroencephalogram along all the electrodes, extremely low amplitude and flattened tracing.

- Absence of a normally functioning cortex on Brainstem auditory evoked response test

5. Evaluation of the patency/ occlusion of the internal carotid and middle cerebral arteries using Digital subtraction and Brain Magnetic Resonance angiography.

The prognosis is poor in children with hydranencephaly. Many of these patients die before the 1st birthday. However, in some cases, they may survive for several years.

CONCLUSIONS:

Treatment is usually palliative. Prognosis is poor in these patients and most have the life span of about 12 months.

Hydranencephaly can be diagnosed in the 2nd trimester antenatal ultrasound. Adequate knowledge and prompt diagnosis of this congenital rare anomaly is important for better decision making and further management. Terminating the pregnancy under eugenic grounds is better when diagnosed early. The criterion for the termination of pregnancy relies on the availability of diagnostic tests to identify conditions incompatible with life or with cognitive dysfunction. Mothers should also be screened with Chromosomal analysis, serology for Cytomegalovirus, Toxoplasmosis and Herpes infections. These help in the counseling of the patient for future pregnancies.

Abbreviations:

CSF – Cerebrospinal fluid, MCA – Middle Cerebral artery, ICA – Internal Carotid artery, VR - Virtual Reconstruction, TORCH – Toxoplasmosis, Cytomegalovirus, Herpes simplex and HIV, HIV – Human immunodeficiency virus.

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