

Scaling New Heights To Diagnose Hydranencephaly

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Abstract: Hydranencephaly is a specific worldwide CNS encephaloclastic disorders characterized by destruction of cerebral hemispheres and the corpus striatum and replacement by a thin membranous sac filled with cerebrospinal fluid, glial tissue and ependymal. The sac represents the relatively intact leptomeninges. In this condition midbrain usually remain safe and intact but ambiguity still persists in mode of transmission. However skull and meninges consistent with normal embryogenesis of the telencephalon show their true identity both in congenital or acquired conditions. In many children at birth impaired vision, infantile spasm or seizures, slow growth, the head appears to be enlarged associated with spasticity and rigidity of arms and legs, thereby both physical and mental retardation seen with poor intellect or skill. The children reveal poor thermoregulation and experience an increased muscular tone (hypertonia) or exaggeration of muscular reflexes (hyperreflexia). Many children attain mortality even prior to delivery or on the onset of birth. MR imaging of the 6 week old female child displays symptoms of irritability, spontaneous reflexes such as, crying, sucking, swallowing and early onset of diabetes insipidus, retarded motor developments,. Diagnosis may be delayed for several months due to relatively normal behaviour, movement of arms and legs, macrocephaly and even normal head size.

I. Introduction

Hydranencephaly is a rare congenital disorder and one of the most severe forms of bilateral cerebral cortical anomaly, in which the cerebral hemispheres are absent and represented by membranous sacs with the remnants of the cortex dispersed over the membranes and replaced by sacs filled with cerebrospinal fluid. The midbrain, cerebellum, thalami, basal ganglia, and choroid plexus are usually not involved. The term hydranencephaly was first introduced by Spielmeyer.

Incidence: 1/10000 live births world wide.

II. Case Report

A single term female baby born via naturalis to a 2nd degree consanguineous married couple with a birth weight of 1.9kg, with no significant family history. Antenatal scan done at 20 weeks of gestational age detected hydranencephaly. Mother was not willing for termination. Baby was delivered via naturalis with uneventful perinatal period. On examination child was lethargic, poor reflexes, microcephaly (<3rd centile) and right upper eye lid coloboma with congenital bilateral club foot. X-ray chest and ECHO reports are unremarkable. CT scan and MRI reports were showing complete absence of cerebral hemispheres and is occupied with cerebrospinal fluid suggestive of hydranencephaly. This baby died on 8th day of life.

III. Mri Findings

- Gross fluid is seen replacing bilateral cerebral hemispheres, basal ganglia and thalami with thinned out bilateral cerebral cortices seen. Frontal and occipital lobes are seen but thinned out. In addition posterior fossa fluid is seen completely replacing inferior cerebellum and portion superior cerebellum. The brain stem is normal. Hydranencephaly with large posterior fossa cyst as described above.
- There is no evidence of intra parenchymal or extra axial haemorrhage.
- No areas of restricted diffusion seen on DWI.
- No mass effect or midline shift.
- No mass lesion noted.
- Visualized orbits and PNS appear normal.
- Fourth ventricle is normal seen not communicating with posterior fossa fluid/cyst.
- Cerebellum– possibly suggestive of large posterior fossa cyst with mass effect on the cerebellar hemisphere in the form of anterolateral displacement.

Imaging features possibly suggestive of hydranencephaly replacing cerebral and with possibly posterior fossa cyst/fluid.

IV. Discussion

Etiopathogenesis of hydranencephaly is still unknown but brain damage is related to early internal carotid artery involvement, so hydranencephaly is categorized as a member of a group of circulatory developmental encephalopathies. Other proposed causes are Intrauterine infections, particularly toxoplasmosis and viral infections (enterovirus, adenovirus, parvovirus, cytomegalic, herpes simplex, Epstein-Barr, and respiratory syncytia viruses) Toxic exposures, such as smoking and cocaine abuse, estrogens and sodium valproate have also been reported. HE has been associated with young maternal age. Most of the case-reports have been diagnosed between the 13th and the 26th week of pregnancy. Some reports may be diagnosed very early in the first trimester. Most affected children die before birth. Those who survive do not initially show evident neurological or clinical signs; archaic reflexes, leg and arms movements are usually normally present at birth, as are sucking and swallowing reflexes. However, more subtle signs, such as feeble crying, difficulty with feeding, hypotonia or wide anterior fontanelle may be present later with severe hypotonia irritability and seizures.

V. Diagnosis

Diagnosis of hydranencephaly can be determined in utero by ultrasonography as early as 12 weeks of gestation. Postnatally Ultrasound, CT scan and transillumination tests are helpful, brain MRI remains the best diagnostic test. Following are differential diagnosis for hydranencephaly.

Prognosis:

Unlike to the unilateral form, the prognosis of hydranencephaly is usually quite poor. Affected patients mostly die in utero. In the survivors, death usually occurs in the first year of Life, However patients with survival of 20, 22 and 32 years have been reported in the literature.

Differential diagnosis of hydranencephaly with hydrocephalus, holoprosencephaly, porencephaly

	Hydranencephaly	Hydrocephalus	Holoprosencephaly	Porencephaly
Head circumference	Normal or slightly smaller	Larger	Normal	Normal
Midline malformations	Absent	Absent	Present	Absent
Brainstem anomalies	Absent	Absent	Present	Absent
Intact cortical rim	Absent	Present	Present	Present
Dilated third ventricle	Absent	Present only in obstructive forms	Absent	Absent
Angiographic investigation	Bilateral internal carotid artery occlusion (not always)	Normal	Normal	Involvement of middle cerebral artery resulting in localized areas, of cortical destruction
Facial malformations	Absent	Absent	Present	Absent
Surgical treatment	Doubt	Useful	Not advisable	Not advisable

Treatment:

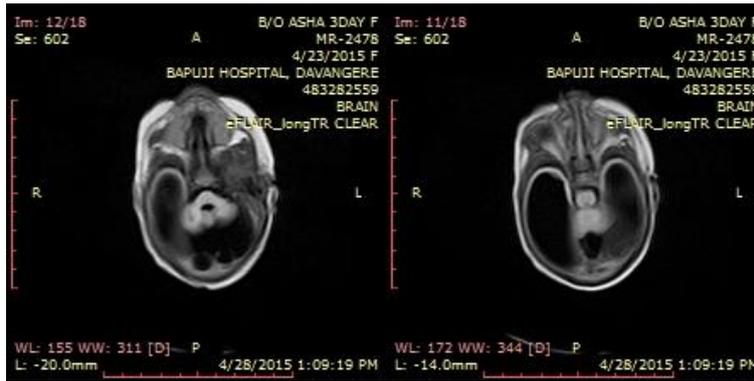
As of date no definitive treatment for hydranencephaly. Symptomatic and supportive therapy remains the treatment.

VI. Conclusion

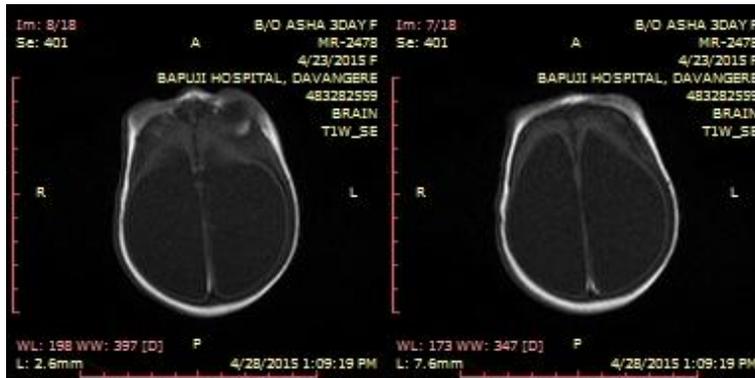
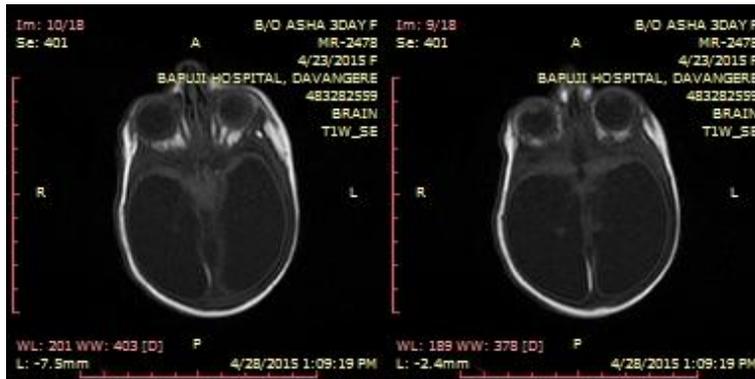
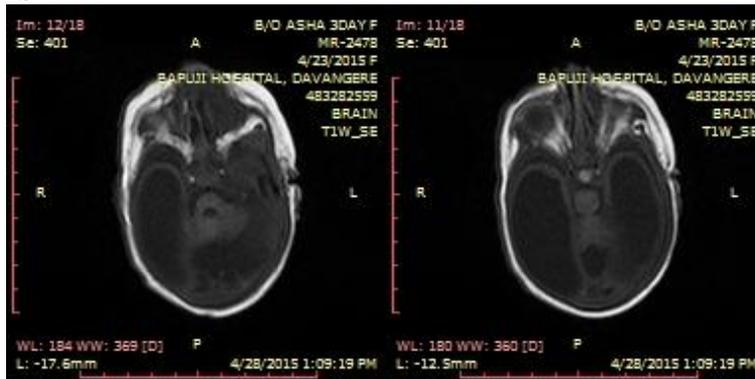
Hydranencephaly is a rare disease with in-utero death as the most typical result and a very poor life expectancy. Most of the children will die before the age of 1 year. As of date no definitive treatment for hydranencephaly. Symptomatic and supportive therapy is remains the treatment. It becomes imperative prior to delivery with the onset of this disorder to show quickness to distinguish this condition from extensive hydrocephalous, holoprosencephaly, large porencephalic cyst and allied variations fair programming relevant to the pertinent treatment can be chalked out. Therefore, in this study causes and cure from poor prognosis to extensive hydrocephalus envisaging the need of improved diagnosis with early shunting procedures have been brought to lime light to deliver the goods.

References

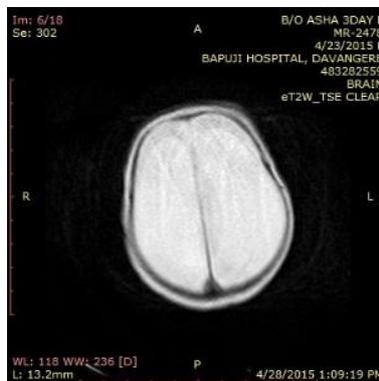
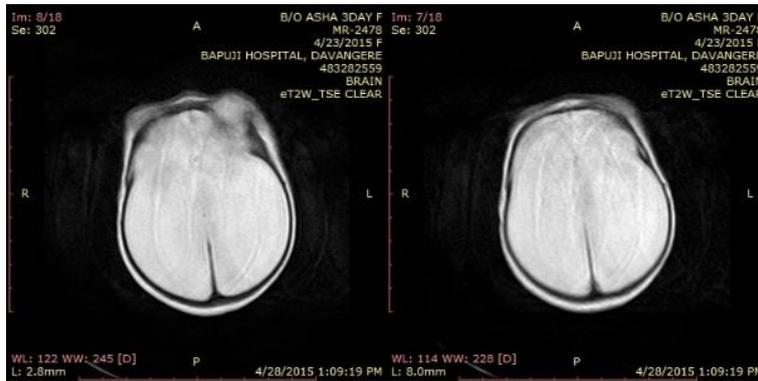
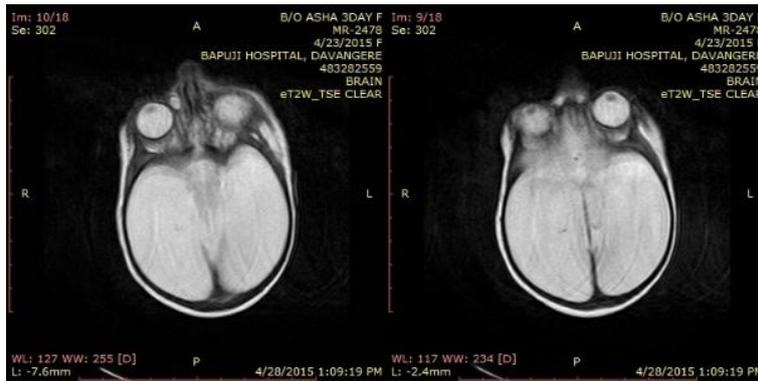
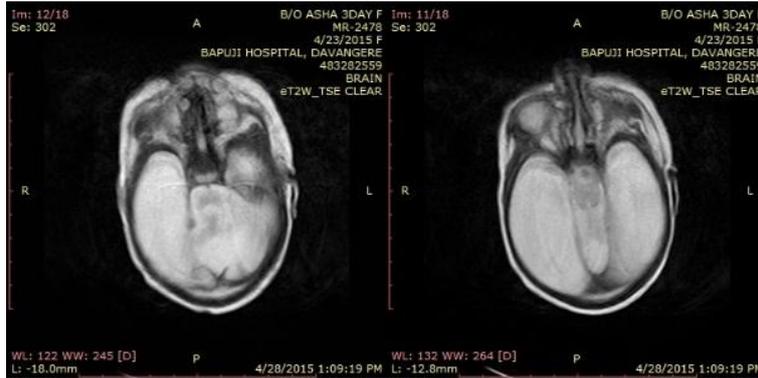
- [1]. Stephen L. Kinsman and Michael V. Johnston: Congenital Anomalies of the Central Nervous System: hydranencephaly. Nelson 20th edition. pg no: 2817.
- [2]. Greco F, Finocchiaro M, Pavone P, Trifiletti RR, Parano E: Hemihydranencephaly: case report and literature review. J Child Neurol 2001, 16: 218-221. Pavone P, Nigro F, Falsaperla R, Greco F, Ruggieri M, Rizzo R, Pratico AD,
- [3]. Pavone L: Hemihydranencephaly: living with half brain dysfunction. Ital J of Pediatr 2013, 39: 3.
- [4]. Spielmeyer W: Einhydranencephales Zwillingpaar. Arch Psychiat Nervenkr 1905, 39: 807.
- [5]. Lin YS, Chang FM, Liu CH: Antenatal detection of hydranencephaly at 12 weeks, menstrual age. J Clin Ultrasound 1992, 20: 62-64.



FLAIR SEQUENCES



T1 SEQUENCES



T2 SEQUENCES



