Kathmandu University Medical Journal (2010), Vol. 8, No. 1, Issue 29, 83-86

Case Note

Hydranencephaly

Pant S[1], Kaur G[2], JK De[3]

[1]Medical Officer, [2]Associate Professor, [3]Professor and Head, Department of Obstetrics and Gynaecology, Manipal College of Medical Sciences

Abstract

Hydranencephaly is a rare congenital condition where the greater portions of the cerebral hemispheres and the corpus striatum are replaced by cerebrospinal fluid and glial tissue. The meninges and the skull are well formed, which is consistent with earlier normal embryogenesis of the telencephalon. Bilateral occlusion of the internal carotid arteries in utero is a potential mechanism. Clinical features include intact brainstem reflexes without evidence of higher cortical activity. The infant's head size and the spontaneous reflexes such as sucking, swallowing, crying, and moving the arms and legs may all seem normal at birth. However, after a few weeks the infant usually becomes irritable and has increased muscle tone and after a few months of life, seizures and hydrocephalus (excessive accumulation of cerebrospinal fluid in the brain) may develop. Other symptoms may include visual impairment, lack of growth, deafness, blindness, spastic quadriparesis (paralysis), and intellectual deficits. Since the early behaviour appears to be relatively normal, the diagnosis may be delayed for months sometimes. There is no definitive treatment for hydranencephaly. The outlook for children with hydranencephaly is generally poor, and many children with this disorder die before their first birthday.

Key words: hydranencephaly, congenital anomaly, vascular disruption, thromboplastin,

A 19 years old, Gravida 2 Para 0 Abortion 1, presented at 31+5 gestational week presented with complaints of pain abdomen and bleeding per vaginum for 1 day. The patient was a booked case in a district hospital and had received routine antenatal care including an anomaly scan at 16 week period of gestation (POG) which had not revealed any abnormality. She gave history of burning micturition at around 24 week POG for which she had taken Nitrofurantoin and urine alkaliniser from the same hospital. Beside this, her first and second trimesters were uneventful.

Past history: History of 1 spontaneous abortion at 20 week POG.

On examination:

Vitals signs:

Pulse-78 bpm ; BP -100/70mm Hg ; Respiratory rate -20/min ; Temp-98.6 F

Abdominal examination: uterus 32 wk size; longitudinal lie, cephalic presentation, fetal heart sound (FHS) @142 bpm, contractions :one every ten minute each lasting for approximately twenty seconds(1/10`/20``).

Routine Antenatal investigations (blood counts, urine routine and microscopy, serology for HIV,HCV and HBV, blood sugars) were normal. Her blood group was B positive.

Ultrasonogram(done at the time of presentation)showed the following findings.

- A single live fetus of calculated gestational age of 32 weeks.
- Right lateral fundal placentation.
- Liquor volume was adequate.
- Massive intracerebral cavitation with non visualisation of cerebral hemispheres.
- Falx,midbrain and posterio fossa structures are visualised suggestive of alobar type of hydranecephaly.

The patient delivered spontaneously, a female baby weighing 1.8 kg. The head circumference was 33.5 cm, length 40 cm and gestational age 32 weeks according to New Ballard Scoring.

Correspondence
Dr. Sadip Pant
Medical Officer
Manipal College of Medical Sciences
E-mail: sadippant@hotmail.com

The baby did not cry for 5 minutes following birth. Oral and nasal suctioning was done, tactile stimulation was given and bag and mask ventilation (BMV) performed. Apgar score at 1 min was 2/10 and at 5 min was 8/10. Baby was shifted to NICU for further management. Head Examination revealed: normal sized brilliantly transilluminant head with wide open posterior fontanelle.

Systemic Examination was within normal limit with intact neonatal reflexes.

The baby was investigated and neurosonogram, echocardiogram and USG abdomen.

The neurosonographic findings were consitent with the obstretic scan findings as mentioned earlier.

Renal and cardiac anomaly ruled out.

The sepsis screen was negative.

The baby was taken against medical advice for economical reasons on 3rd postpartum day.

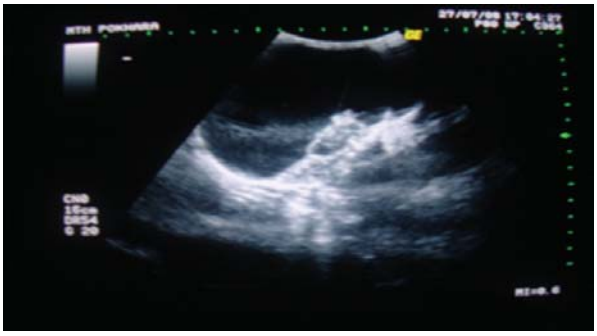

Fig 1: Ultrasonogram obtained at 31+5 weeks shows unfused thalami surrounded by fluid in this fetus with hydranencephaly. Note the lack of cerebral hemispheric tissue.

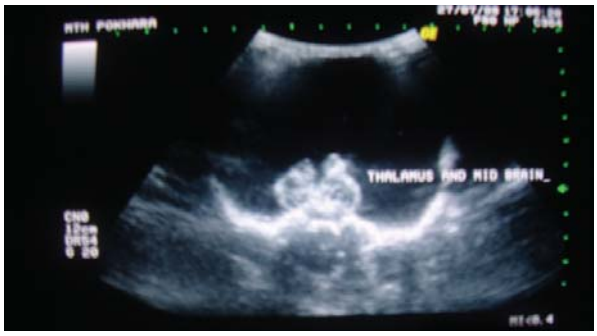

Fig 2: Sagittal image in the same patient as in Image 1 again demonstrates the absence of the cerebral hemispheres (which have been replaced by fluid). Because of the early age of gestation, hydranencephaly was difficult to distinguish from hydrocephalus.

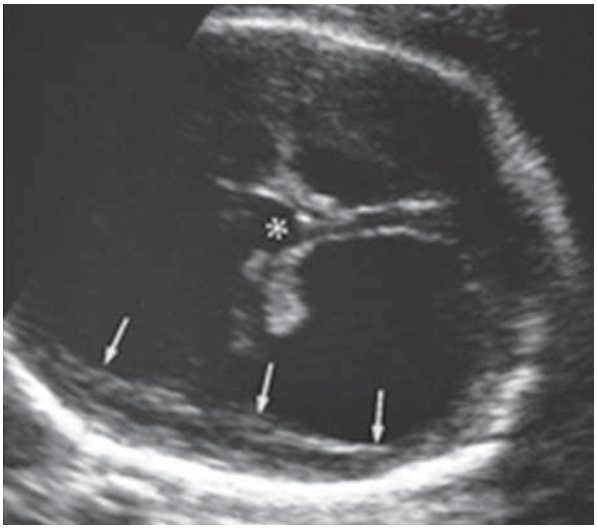

Fig 3: Hydrocephalus. A transaxial USG scan of a fetus at 30 weeks gestation demonstrates an enlarged fetal head with thinned but present temporoparietal cortical mantle (arrows) along the posterolateral aspect of the calvaria. Although the temporoparietal cortical mantle is also present anteriorly, it cannot be appreciated because of reverberation artifacts[14].

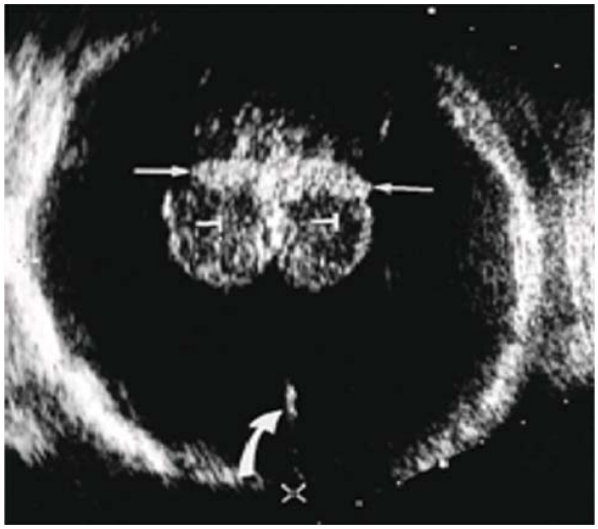

Fig 4: Hydranencephaly.Transaxial image near the vertex demonstrates a discontinuous falx midline echo (curved arrow). Normal hyperechoic choroid plexuses (straight arrows) are seen posterior to the thalami.There is no identifiable cortical mantel[14].

Discussion

Hydranencephaly is a rare, isolated abnormality occurring in less than 1 per 10,000 births worldwide [1–3]. It is the most severe form of bilateral cerebral cortical destruction. This is a condition in which the cerebral hemispheres are absent and replaced by sacs filled with cerebrospinal fluid. Hydranencephaly occurs after the brain and ventricles have fully formed, usually in the second trimester.

Etiopathogenesis

Five theories have been proposed:

1) Bilateral occlusion of the supraclinoid segment of the internal carotid arteries or of the middle cerebral arteries[4].
2) An extreme form of leukomalacia formed by confluence of multiple cystic cavities[5].
3) Diffuse hypoxic-ischemic brain necrosis[6].
4) Infection causing necrotizing vasculitis. Intrauterine infections, particularly toxoplasmosis and viral infections (enterovirus, adenovirus, parvovirus, cytomegalic, herpes simplex, Epstein-Barr, and respiratory syncytial viruses), have been implicated in a number of cases[7,8]. Toxic exposures and cocaine abuse have been reported, and hydranencephaly has been described in rare syndromes[9].
5) Thromboplastic material from a deceased co-twin1[10].

There is liquefaction of the brain tissue in the area involved (usually the hemispheres),with replacement of the neural tissue by cerebrospinal fluid and preservation of the membranes (the meninges).

Presentation

An infant with hydranencephaly may appear normal at birth or may have some distortion of the skull and upper facial features due to fluid pressure inside the skull. The infant's head size and spontaneous reflexes such as sucking, swallowing, crying, and moving the arms and legs may all seem normal, depending on the severity of the condition. However, after a few weeks the infant usually becomes irritable and has increased muscle tone (hypertonia).After several months of life, seizures and hydrocephalus may develop. Other symptoms may include visual impairment, lack of growth, deafness, blindness, spastic quadriparesis/plegia and intellectual deficits. Some infants may have additional abnormalities at birth including seizures, myoclonus (involuntary sudden, rapid jerks), and respiratory problems.

Diagnosis

Diagnosis may be delayed for several months because the infant's early behavior appears to be relatively normal. Transillumination, an examination in which light is passed through body tissues, usually confirms the diagnosis.

Preliminary diagnosis may be made in utero via standard ultrasound, and can be confirmed with a level II or higher ultrasound.

Prognosis

There is no standard treatment for hydranencephaly. Treatment is symptomatic and supportive. Hydrocephalus may be treated with a shunt (e.g.a ventriculoperitoneal shunt). The prognosis for children with hydranencephaly is generally quite poor. Death usually occurs in the first year of life[11].

The differential diagnosis

The common differentials for hydranencephaly include extreme hydrocephalus, alobar holoprosencephaly and porencephaly. On ultrasound, hydrancephaly presents as a large cystic mass filling the entire cranial cavity with absence or discontinuity of the cerebral cortex and of the midline echo. The appearance of the thalami and brainstem protruding inside a cystic cavity is characteristic. With either extreme hydrocephaly, alobar holoprosencephaly or porencephaly, these structures should still be surrounded by a rim of cortex, and the choroid plexuses should be normally visible. Magnetic Resonance Imaging study can aid in confirmation of these findings. There are important reasons to differentiate **hydranencephaly** from hydrocephalus; these reasons relate to prognosis and management[12]. Sutton and associates[13] followed 10 neonates with serial computed tomography, electroencephalograms, and developmental evaluations for 4-23 months. Two syndromes were defined. The five infants with hydranencephaly demonstrated neither neurologic nor radiologic improvement beyond 1 month of age despite aggressive surgical management and shunt placement. The five infants with maximal hydrocephalus improved dramatically over time following shunt placement.

Conclusion

Although the number of cases of hydranencephaly diagnosed in utero by ultrasonography can be counted in fingers[15], the importance of such a diagnosis cannot be overlooked. The timely diagnosis of a congenital anomaly which is incompatible with postnatal life like hydranencephaly can at least help in preventing maternal morbidity (in medical, psychological and economical terms) by contemplation of medical termination of pregnancy under eugenic grounds. In addition, the termination of pregnancy as late as the third trimester may be justified when an antenatal diagnosis of hydranencephaly is made.The criteria for termination includes the availability of reliable diagnostic tests that can accurately predict a condition that is either

incompatible with post-natal life or characterized by the absence of cognitive function[16]. Nevertheless, this was inappropriate in this particular case as the parturient had already presented in labour. Had the anomaly been diagnosed electively rather than incidentally also, the questions of bioehics and legal issues would have influenced the obstretician's response to abortion. This demands the dire need for addressing this issue in the national medical termination of pregnancy guidelines and possibly giving it a space in it so that the moral obligations of a doctor is not overwhelmed by bioethics and legal issues in such cases. If termination of pregnancy is contemplated, chromosomal analysis, serology for *CMV*, *toxoplasmosis*, and *Herpes* cultures should be obtained as these findings may aid in counselling for future pregnancies[17].

References

1. Filly RA. Ultrasound evaluation of the fetal neural axis. In: Callen PW,editor. Ultrasonography in obstetrics and gynecology. 3rd ed. Philadelphia, Pa: Saunders; 1994.p.199-218.
2. Nyberg DA, Pretorius DH. Cerebral malformations. In: Nyberg DA, Mahony BS, Pretorius DH, eds. Diagnostic ultrasound of fetal anomalies: text and atlas. Chicago, Ill: Year Book Medical; 1990.p.98-121.
3. Chervenak FA, Isaacson GC,Campbell S. Hydranencephaly. In:Chervenak FA, Isaacson GC, Campbell S, eds. Ultrasound in obstetrics and gynecology. Boston, Mass : Little, Brown; 1993. p.1187-8.
4. Myers R. Brain pathology following fetal vascular occlusion: an experimental study. Invest Ophthalmol. 1969;8:41.
5. Nahmias A, Keyserling H, Kerrick G, et al. In: Diseases of the fetus and newborn infant. Philadelphia:WB Saunders ;1983.p.636-78.
6. Hoyme HE, Higginbottom MC, Jones KL.Vascular etiology of disruptive structural defects in monozygotic twins. Pediatrics. 1981;67:288-291.
7. Hutto C, Arvin A, Jacobs R,et al. Intrauterine herpes simplex infections. J Pediatrics. 1987;110:97-101.
8. Christie J, Rakusan T, Martinez M, et al. Hydranencephaly caused by congenital infection with herpes simplex virus. Pediatric Inf Disease. 1986;5:473-8.
9. Rais-Bahrami K, Naqvi M. Hydranencephaly and maternal cocaine use: a case report. Clin Pediatr. 1990; 29:729-30.
10. Larroche J, Droulle P, Dalezoide A, et al. Brain damage in monozygous twins. Biol Neonate. 1990;57:261-78.
11. Hadi H, Mashini I, Devoe L, et al.Ultrasonic prenatal diagnosis of hydranencephaly. A case report. J Reprod Med. 1986;31:254-6.
12. Nicolaides KH, Snijders RJM, Gosden CM, Berry C, Campbell S. Ultrasonographically detectable markers of fetal chromosomal abnormalities. Lancet. 1992; 340:704-7.
13. Sutton LN, Bruce DA, Schut L. Hydranencephaly versus maximal hydrocephalus: an important clinical distinction. Neurosurgery. 1980; 6:34-8.
14. Kurtz AB, Johnson PT. Diagnosis please. Case 7: Hydranencephaly. Radiology. 1999;210(2):419-22.
15. H Hidalgo, J Bowie, ER Rosenberg, PC Ram, K Ford, and E Lipsit.In utero sonographic diagnosis of fetal cerebral anomalies. American Journal of Roentgenology. 1982; 39(1):143-8.
16. Chervenak F, Farley M, Walters L. When is termination of pregnancy during the third trimester morally justifiable? N Engl J Med. 1984;310:501.
17. Bordarier C,Robain D.Familial occurrence of prenatal encephaloclastic damage. Anatomicoclinical report of 2 cases. Neuropediatrics. 1989:103-6.