Hydranencephaly: A rare case report

Nalini Sharma1, Vinayak Jante2, Rituparna Das1, Subrat Panda3, Manddeep Sagar4

From 1Assistant Professor, 2Senior Resident, 1Associate Professor, Department of OBG, 3Senior Resident, Department of Radiology, North Eastern Indira Gandhi Regional Institute of Health and Medical Sciences, Shillong, Meghalaya, India

ABSTRACT

Hydranencephaly (HE) is a rare condition occurring in <1/10,000 births worldwide. It is one of the recognized forms of brain malformations that are usually associated with intrauterine fetal demise rarely seen in postnatal life. HE can often be misdiagnosed due to certain common features with other neurological abnormalities such as hydrocephalus, holoprosencephaly, and porencephaly. Here, we report the case of a 26-year-old pregnant patient at 34 weeks who was referred with ultrasonography finding of HE which was confirmed by fetal MRI. The decision to deliver the baby was taken expecting an extremely poor outcome after discussing with the family.

Key words: Cerebrospinal fluid, Hydranencephaly, Hydrocephalus, Internal carotid artery occlusion

Hydranencephaly is a rare condition occurring in <1/10,000 births worldwide. It is an isolated abnormality with a severe prognosis [1]. It is one of the most severe forms of bilateral cerebral cortical anomaly, affecting more than one-third of all children. In this condition, the cerebral hemispheres are replaced by a membranous sac filled with cerebrospinal fluid, glial tissue, and ependyma. However, however, there will be the preservation of the skull. There is variable involvement of the inferior frontal, temporal, and occipital lobes. The midbrain, cerebellum, thalami, and basal ganglia are usually not involved. HE can affect only one hemisphere, which is called hemihydranencephaly, which is even rarer than HE and may have a better prognosis [2]. HE is one of the recognized forms of brain malformations usually associated with intrauterine fetal demise rarely seen in postnatal life. It can often be misdiagnosed due to certain common features with other neurological abnormalities such as hydrocephalus, holoprosencephaly, and porencephaly [2]. The exact etiology is not known. The most common cause is found to be occlusion of the supra-clinoid part of the internal carotid artery (ICA), which leads to infarction of cerebral structures [3]. So far, no sex or racial preference has been discovered [1].

The rationale of our case report is to report on this condition, utilizing data from the literature, and also present our experience with a pregnant patient who presented in her third trimester with bilateral fetal HE.

CASE REPORT

A 26-year-old Primigravida at 34 weeks of gestation was referred to the Department of Obstetrics and Gynecology of our institute. In view of pre-eclampsia and bilateral fetal HE, her marriage was non-consanguineous. There was no family history of genetic or congenital anomalies. She did not have a regular antenatal check-up and did not perform routine appointment scans, anomaly scans, or growth scans on the fetus during pregnancy. She had her first antenatal visit in the third trimester at 33 weeks at a private hospital where she was diagnosed with pre-eclampsia and on antenatal ultrasound, the fetus was found to have bilateral HE. She was referred in view of the same condition to our institute for a second opinion.

A physical examination at admission revealed blood pressure of 150/102 mmHg, heart rate of 92 beats/min, and respiratory rate of 20 breaths/min. Ophthalmic and other systemic (central nervous system, cardiovascular system) examinations were within normal limits.

Initial laboratory tests showed normal hemoglobin (13 g/dL) and platelet count (200 × 109/L). The evaluation of coagulation parameters was within normal limits. Her biochemical analysis for renal and hepatic function showed total protein (5.60 g/dL), albumin (2.8 g/dL), albumin/globulin ratio (1.14), proteinuria (++), normal transaminases (ALT 23.2 IU/L, AST 29.2 IU/L), and lactate dehydrogenase (LDH 233 IU/L). Blood pressure was well controlled with the tablet Labetalol 100 mg twice a day.

Obstetric ultrasound was repeated and the findings corresponded with the previous ultrasound that is bilateral HE.
Although ultrasound is good enough for the diagnosis of HE, on patients’ request to confirm the diagnosis and also to differentiate between other close differentials such as hydrocephaly and holoprosencephaly, fetal Magnetic resonance imaging (MRI) was done, which revealed fetal HE and deformed posterior structures (Fig. 1).

After consulting with the family, the decision was made to deliver the baby at 34 weeks and 5 days, with the expectation of an extremely poor outcome. She was planned for induction of labor with a pharmacological method. The duration of induction was prolonged, requiring 6 doses of 50 microgram Misoprostol given at a 6-h interval. She delivered a female fresh stillborn baby of 1.9 kg and had a normal physical appearance. The head of the baby was of normal size (head circumference 32 cm). The postpartum period was uneventful and blood pressure was controlled with tablets of Labetalol 100 mg twice a day, and the patient was discharged on day 3.

DISCUSSION

HE is characterized by the absence of cerebral hemispheres which are replaced by cerebrospinal fluid and necrotic debris, covered by leptomeninges. It is an encephaloclastic abnormality. The midbrain, thalamus, basal ganglia may be preserved, and the flax cerebri is usually present within the skull [3].

The exact etiopathogenesis of HE is still not known, so several theories have been postulated. However, the most accepted theory as suggested by many researchers is brain damage caused by early involvement of ICAs which is suggested by i) angiographic and autopic observations where both aplastic and hypoplastic arteries were reported in ICAs and ii) the anatomic distribution follows the ICA supply [2]. Mayers did an experiment on monkeys to describe the etiology of HE. In his study, monkey fetuses at different gestational ages were subjected to ligation of the bilateral ICAs and jugular veins. The fetuses were then restored to the uterus and delivered at term. An examination of baby monkey brains revealed HE due to vascular shutdown predominantly when carried out during earlier gestational age.

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is advisable to help them to be prepared for the potential outcome through proper counseling of the parents regarding the poor prognosis and potential management options [3]. The termination of the pregnancy is recommended once a definitive diagnosis is made due to a poor prognosis. In cases of macrocrania, the term cephalocentesis can be suggested [10].

CONCLUSION

The findings we described led to the diagnosis of HE, which was confirmed by fetal MRI. Postnatal CT/MRI will be more appropriate, but in our case, it was a stillbirth, so postnatal investigations could not be done. It is critical to diagnose HE early so that obstetric complications can be avoided and, most importantly, proper parental counseling can be provided. Our case was an unbooked case as she did not have a regular antenatal check-up, so the diagnosis was made in the third trimester. It is challenging for doctors to manage such cases in the late trimester and also very distressful for the family. It is important for preparing the optimal conditions for delivery and allowing for specialized care by pediatricians by early diagnosis of HE.

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