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CASE REPORT

A rare variation of hydranencephaly: case report [version 1; peer review: 1 approved, 1 approved with reservations]

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Abstract

Hydranencephaly is a rare severe abnormality characterized by replacement of cerebral hemisphere with fluid covered by leptomeninges such that absent cerebral cortex. We present the Ultrasonographic diagnosis of a case of fetal Hydranencephaly at 38 weeks of gestation. Sonography revealed the absence of cerebral cortex, thalami and basal ganglia with disrupted falx and preserved posterior fossa structures. This was the first reported case of Hydranencephaly with absence thalami and basal ganglia along with midbrain. Confirmation of diagnosis was made with postnatal computed tomography. Even though it was diagnosed in the late third trimester, it allowed prompt and finest obstetric management which reduced the risk for both fetus and mother. Furthermore it allowed arrangement of optimal conditions for birth at a unit with accessible specialized pediatric facilities.

Open Peer Review

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 School of Medicine, Baltimore, MD, USA

2. **Athanasios Petridis**, University of Schleswig-Holstein, Kiel, Germany

Any reports and responses or comments on the article can be found at the end of the article.

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Editorial note:

Please note that the refereeing status of this version was changed from “indexed” to “[v1; ref status: approved 1, approved with reservations 1]”.

When Version 1 of this article was first published, *F1000Research* was still in its beta phase; during this period articles that received any two of “Approved” or “Approved with Reservations” statuses from the reviewers were labelled as “indexed”. When the journal was formally launched in January 2013, the requirements for indexing were tightened, and now only articles that are given either two “Approved” or one “Approved” plus two “Approved with Reservations” statuses by the reviewers are labelled “indexed”.

Later version(s) of this article received the necessary approval status from the reviewers and are “indexed” according to the new criteria.

Introduction

Hydranencephaly is a rare abnormality in which the cerebral hemispheres are almost absent and have been replaced with fluid. It was first described by crueveilhaer (1892) as “Anencephalie hydrocephalique” or “Hydroanencephalie”¹. Crome and Sylvester reviewed the disease and defined it as a congenital condition². It is a very rare isolated abnormality occurring less than 1 per 10,000 births world wide³. It is said to be present in 0.2% of infant autopsies and approximately 1% of babies are diagnosed clinically as hydrocephalus⁴. It’s important to distinguish early between Hydranencephaly and extreme hydrocephalus, because the latter carries a considerably superior prognosis⁵. We report a rare case of Hydranencephaly which was diagnosed in late third trimester followed by successful obstetric management.

Case presentation

A 28-year-old woman, gravida 2 para 1 was presented to our tertiary care institution at 38 weeks’ gestation because of lower abdominal pain. She and her partner were Sinhalese and unrelated. There was no family history of genetic or congenital anomalies. Her first pregnancy was uncomplicated normal vaginal delivery with normal healthy male child. She had not attended the local antenatal clinic regularly where she was recognized as a noncompliant patient. She had not undergone routine dating scan, anomaly scan or the growth scan of the fetus. She was a nonsmoker and there was no history suggestive of congenital infections or exposure to toxins. The clinical obstetric examination was unremarkable. The cardiotocography of the fetus was normal.

Obstetric ultrasound scan showed fluid filled cranial cavity with absent cerebral cortex, thalami and basal ganglia. The third ventricle dilated and remnants of midbrain structures were seen (Figure 1). The cerebellum and other posterior fossa structures were preserved. The falx cerebri was disrupted (Figure 2). There was no polyhydramnios. Umbilical artery Doppler studies were normal. Sonographically it was suggestive of Hydranencephaly. Her blood group was B positive and Infectious disease Antibody test showed negative titers for Rubella, HIV, Hepatitis B and Toxoplasmosis. We planed caesarian section to avoid obstetric complications due to possible cephalopelvic disproportion and minimize the risk for both mother and fetus. Specialized Pediatric team was pre

Figure 1. Prenatal ultrasound scans image of the fetus. Transabdominal transverse section of the fetal head at 38 weeks of gestation shows dilated third ventricle (V) with absent thalami and basal ganglia and cerebral cortex.

Figure 2. Prenatal ultrasound scans image of the fetus. Transabdominal transverse section of the fetal head shows disrupted falx (F).

informed. Parents were counseled regarding poor prognosis of the newborn’s condition. At 39 week of gestation emergency caesarian section performed due to fetal distress. The newborn was a 2,980g girl with normal physical appearance and normal sized skull (head circumference = 33.5 cm) which was brilliantly transilluminated.

Newborn was transferred to neonatal Intensive care unit because of the respiratory distress. Four days after delivery Computed tomography (CT) of the newborn’s head was performed without intravenous contrast. On CT there was no cerebral cortex, thalami

or basal ganglia identified. The third ventricle dilated and remnants of midbrain structures were seen (Figure 3). The cerebellum and other posterior fossa structures were preserved with disrupted falx cerebri (Figure 4a, 4b).

The CT scan confirmed the diagnosis Hydranencephaly. Baby died after two weeks of birth due to cardiac arrest. The parents declined a postmortem examination.

Discussion

Hydranencephaly is an encephaloclastic abnormality characterized by replacement of cerebral hemisphere with the cerebrospinal fluid

Figure 3. CT image of 2 days old newborns brain. Dilated third ventricle (V) with absent thalami and basal ganglia and cerebral cortex.

Figure 4. CT images of 2 days old newborns brain. (a) Intact Cerebellum (C) and other posterior fossa structures. (b) Shows Disrupted Falx cerebri (F).

and necrotic debris surrounded by leptomeninges such that no cerebral cortex is present but there may be partial preservation of portion of the occipital lobe⁶. The midbrain, thalamus, basal ganglia, choroid plexuses, cerebellum and brain stem are usually preserved and contained within normal skull. The Falx cerebri is usually present but may be partially or completely absent. The septum pellucidum may be absent.

The aetiopathogenesis of Hydranencephaly is heterogeneous, but several theories have been postulated. It has been suggested that bilateral occlusion of supraclenoid segment of the internal carotid artery or middle cerebral⁷ arteries before the 24 weeks of gestation cause ischemia, edema, autolysis and disappearance of cerebral hemispheres respectively⁸. Nevertheless some reports suggest occlusion of arteries due to temporary spasm or compression rather than direct occlusion⁹. Intrauterine infections also cause necrotizing vasculitis or local destruction of brain. Infections like congenital toxoplasmosis or viral infections (Adenovirus, cytomegalovirus, Enterovirus, Epstein-Barr virus, herpes simplex virus, Parvovirus, and respiratory syncytial viruses) have been implicated in numerous cases³. Fetal hypoxia due to exposure to toxin like Carbon monoxide or butane gas during antenatal period may result diffuse hypoxic ischemic brain necrosis lead to Hydranencephaly¹⁰. Thrombotic material from a deceased co-twin in monochorionic twin pregnancies reported to be implicated¹¹. Hydranencephaly has been described in rare syndromes¹².

Ultrasonographic findings include large cystic mass filling entire intracranial cavity with absence of cerebral cortex. The head size may be normal or large¹³. Appearance of thalami and brainstem protruding in to cystic cavity is characteristic, together with a midline echo from the remnant of falx, the tentorium cerebella and cerebellum¹⁴. The third ventricle and choroid plexuses are often visible. Absence of septum pellucidum may give rise to what appears to be a single ventricle in midline¹⁵. The major differential diagnosis includes extreme hydrocephalus, alobar holoprosencephaly and porencephaly. In these conditions the above mentioned structures will still be surrounded by rim of cortex. In extreme hydrocephalus, the thin cortical mantle may be hard to identify sonographically and Magnetic resonance imaging (MRI) or rarely intrauterine CT scan might aid diagnosis¹³. The ultrasound is the best diagnostic tool during the prenatal period and postnatal period it's MRI¹⁶⁻¹⁸ or CT. It is important to distinguish Hydranencephaly from other differential diagnosis because they carry better prognosis. The most of the cases detected second half of pregnancy¹² and there have been reported cases of Sonographic evaluation of fetal Hydranencephaly in the first trimester¹⁹.

Hydranencephaly has an irretrievably poor prognosis, with merely remaining brainstem functions. Some die at birth, but most infant die within the first year of life²⁰ and if survived they are profoundly retarded. The recurrence risk is negligible¹⁴. Because of the poor prognosis, termination of pregnancy is recommended once definitive diagnosis has been established¹³. If macrocrania is found in late pregnancy cephalocentesis may be indicated (aid delivery)¹³. Counsel parent regarding poor prognosis and management options are very important.

Ultrasonographic findings, in the case we have described, made a diagnosis of Hydranencephaly particularly likely and this was confirmed by postnatal CT of the fetal head. This is the first published case of Hydranencephaly with absent thalami and basal ganglia along with midbrain. Sonographic evaluation is sufficient for the prenatal diagnosis of Hydranencephaly in most cases, and MRI or intrauterine CT cannot be considered a first-line diagnostic tool.

Conclusions

Early diagnosis is important as an early treatment options avoids obstetrics complications and it may be very useful for giving appropriate advice to parents during the pregnancy and also for preparing the optimal conditions of birth at a unit with available specialized pediatric facilities.

Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editors.

Author contributions

SKR and BTBW reviewed the manuscript. BTBW and GKR were involved in drafting the manuscript and reviewing the literature. BTBW was a major contributor in revising the manuscript and getting informed consent from patient. GKR, SKR and BTBW were involved in reviewing the literature. All authors were responsible for the diagnosis, treatment and follow-up of the patient. All authors read and approved the final manuscript.

Competing interests

No competing interests were disclosed.

Grant information

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Athanasios Petridis

Department of Neurosurgery, University of Schleswig-Holstein, Kiel, Germany

This case report is interesting. Except of some grammatical mistakes, I do have one suggestion.

The authors should also discuss the social aspects and the social stress which accompanies families with hydranencephalic children. It is important to induce abortion when possible in such cases since the prognosis is very poor. Early diagnosis is the most important aspect in such cases. The paper of mine, [Petridis et al. \(2011\)](#) deals with the social aspects of hydranencephaly and should be cited by the author.

Competing Interests: No competing interests were disclosed.

I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard, however I have significant reservations, as outlined above.

Author Response 31 Jan 2013

Buddhika Wijerathne, Teaching Hospital Peradeniya, Peradeniya, Sri Lanka

We would like to thank Dr Petridis for the valuable time spent reviewing our manuscript and the important comments he has made. We have attempted to address and incorporate most of the concerns that were raised in version 2 of our article.

Competing Interests: No competing interests were disclosed

Reviewer Report 12 November 2012

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Jeffrey Chinsky

Department of Pediatrics, Johns Hopkins University School of Medicine, Baltimore, MD, USA

This case report presents an interesting case of apparent hydranencephalic changes, in addition to more extensive developmental brain anomalies (lack of thalamus and basal ganglia). However, including the points made below would make the manuscript much more useful as an educational resource:

The manuscript would be enhanced if the author's did more than just itemize a list of associated etiologies for hydranencephaly. The authors should provide a paragraph explaining how any of these etiologies could specifically produce the anomalies observed (cerebral arterial stroke or spasm of which specific vessels) and perhaps contrast it with any syndromic or associated genomic alterations listings of similar brain findings (or at least provide evidence that they researched this aspect).

I'm not sure all would accept this as a variant of the same processes that cause classic hydranencephaly since it appears to be so much more extensive anomalous formation of the final fetal brain. The authors may have to do a little more research and discussion to support their contention based on how others who have written about hydranencephaly and define the condition.

There are several grammatical errors (e.g. in the Abstract "such that absent cortex" is an incomplete phrase to end the sentence) which should be addressed, and I suggest the article is proof read again to ensure it reads how a published manuscript should do.

Competing Interests: No competing interests were disclosed.

I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard.

Author Response 31 Jan 2013

Buddhika Wijerathne, Teaching Hospital Peradeniya, Peradeniya, Sri Lanka

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Competing Interests: No competing interests were disclosed

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