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Case Report

Hydranencephaly: A Rare Cause of Macrocephaly

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Received: September 22, 2023; **Published:** December 13, 2023

Abstract

Hydranencephaly is an encephaloclastic anomaly characterised by the absence of the cerebral hemispheres, which are replaced by a large, fluid-filled cavity. The differential diagnoses of hydranencephaly are severe hydrocephalus and holoprosencephaly. It is usually identified prenatally via ultrasound, but can also be diagnosed postnatally. The prognosis is very poor. We present a clinical case of this rare disease in a 3 days old male infant. We diagnosed it through the evaluation of clinical features and brain CT. A brief review of the related literatures will be reviewed through this article.

Keywords: *Hydranencephaly; Macrocephaly; Congenital Brain Malformation; Imaging; CT*

Introduction

The word hydranencephaly is a fusion of hydrocephalus and anencephaly, but the condition actually represents a distinct disorder. It is characterized by the disappearance of most of the cerebral hemispheres, which are replaced by a cyst bordered by a thin membrane limited on its external face by the leptomeninge [1]. The prognosis is universally poor, and most newborns die in the first year [2]. Imaging, in particular CT and ultrasound, offer a unique diagnostic tool for the evaluation of these infants. In this report, we discuss the effect of imaging on the diagnostic work-up of hydranencephaly.

Case Report

A newborn admitted at 3 days of age, male, from a monitored pregnancy estimated at 37 weeks of amenorrhea, no notion of consanguinity, delivered by C-section of a 38 year old mother. Since birth, the newborn presented major macrocrania, early vomiting and refusal to suckle in a context of apyrexia. Clinical examination found a hypotonic subicteric newborn with a sunset gaze. The initial head circumference was 51 cm. Cranial ultrasound shows the majority of the skull filled with fluid. Cerebral tissue is seen centrally and posteriorly corresponding to thalami, occipital lobes and cerebellum. No frontal or parietal lobes are identifiable. Falx is noted in the midline, and it appears normal.

Non contrast CT demonstrates marked enlargement of the skull with no anterior circulation cerebral cortex identified. Parts of the occipital lobes, the thalami and the posterior fossa appear preserved. The falx is present (Figure 1). The diagnosis of hydranencephaly was made. Surgery was not indicated, nor an external shunt. Palliative care was proposed at day 17 of life at home with psychological family therapy was considered for the parents and the close of kin.

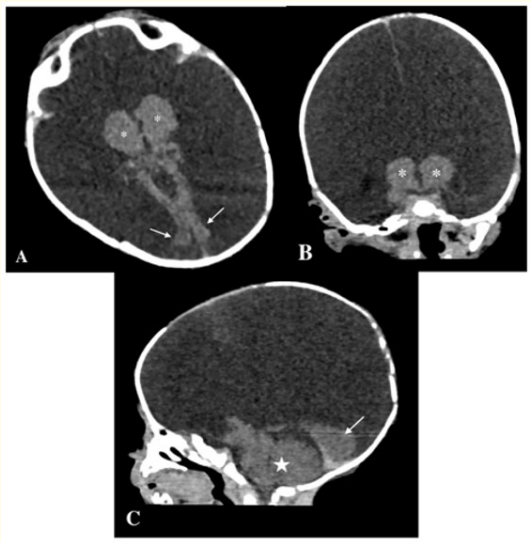

Figure 1: *Axial (A), Coronal (B) and Sagittal (C) non-contrast computed tomography (CT) scans show no remaining cortical tissue, a preserved occipital lobes (arrow), thalami (asterisks), and posterior fossa (star). The falx is present.*

Discussion

Hydranencephaly is a rare congenital brain malformation. The frequency of central nervous system malformations is estimated to be between 5 and 10 per 1000 births [1]. Hydranencephaly was fully described as a different entity from hydrocephalus in 1972 by Crome [1]. It is characterized by the disappearance of most of the cerebral hemispheres. The fluid-filled sac that replaces the brain is formed by the leptomeninges and the cerebrospinal fluid. Its incidence is less than 1 case per 10,000 births [4].

The causes of hydranencephaly are not yet clear. The hypothesis most often described in the literature is bilateral occlusion of the supra-clinoid segment of the internal carotid arteries [5]. This hypothesis could explain hydranencephalies with bilateral absence of cerebral hemispheres but with presence of subcortical tissue and subtentorial parenchyma that is dependent on posterior cerebral circulation. The second hypothesis is intrauterine infection. The germs involved are essentially: cytomegalovirus, varicella, rubella, and less frequently, toxoplasma, herpes and syphilis. The infection results in multi-visceral involvement. The type of brain damage depends on when the fetus was infected. During the first four weeks of infection, cytolysis and vasculitis lesions occur. These are responsible for the constitution, during the following weeks, of areas of necrosis and sclerosis, thrombosis and micro-glial nodules. This results in a more or less extensive destruction of the brain tissue. The other hypothesis is fetal hypoxia due to maternal exposure to carbon monoxide or butane gas leading to massive tissue necrosis with cavitation and resorption of the necrotic tissue [6].

Antenatal diagnosis can be made by ultrasound as early as the 13th week of pregnancy.

The sonographic appearances of hydranencephaly during the early stages of disease would be consistent with acute destruction of the cerebral cortex, giving it a characteristic homogeneously echogenic pattern within the cranial cavity representing blood and necrotic debris. Over time, this content is progressively replaced by more anechoic fluid as the result of progressive liquefaction of blood clots and brain tissue and continued production of cerebrospinal fluid by the choroid plexuses, leading to the classic sonographic appearance of

hydranencephaly as seen at the end stages of disease [7]. The falx cerebri may be incomplete; however, it is usually present and can help to distinguish hydranencephaly from alobar holoprosencephaly [8]. Clinically at birth, hydranencephaly usually presents with normocephaly rather than macrocephaly but occasionally may even be associated with microcephaly [9]. Cranial ultrasonography shows enlargement of fluid spaces with little if any overlying cerebral parenchyma except for the posterior fossa and para medial location along the falx [10]. Our patient underwent a cranial ultrasound which concluded that the brain parenchyma was absent and that the thalami and the posterior cerebral fossa were preserved.

CT and MRI allow to observe the replacement of the brain parenchyma by the fluid. Most often there is an absence of residual cortical tissue. Islands of preserved residual tissue may be found in the occipital poles and orbitofrontal regions. Medial temporal tissue may be seen, as the medial temporal lobes are supplied by the basilar circulation. The thalamus and posterior cerebral fossa are preserved. The falx is usually present [11]. The CT appearance of hydranencephaly may be simulated by other entities, such as hydrocephalus, alobar holoprosencephaly, or severe postinfarctive entities. In alobar holoprosencephaly there are usually coexisting midline malformations, no falx is observed, and the residual cortex has a pancake morphology. Ventricular structures can be appreciated by CT in cases of severe anoxia and severe bilateral porencephaly. Close inspection of scans usually reveals the presence of some cortical mantle in severe hydrocephalus. This mantle is absent in classic cases of hydranencephaly [9]. Anterior and middle cerebral arteries are not visualized in hydranencephaly, and color Doppler may be useful for differential diagnosis [11].

The prognosis of HE is usually poor. The observed survival is due to the preservation of subcortical and brainstem regions that contain the neural circuitry necessary to maintain body temperature, blood pressure, cardiorespiratory and other vital functions. While most patients do not survive beyond the neonatal stage. The life span rarely exceeds the neonatal period. The treatment depends on palliative care and psychological support for the parents [12].

Conclusion

Hydranencephaly is a rare but serious cerebral malformation that poses a problem of differential diagnosis with hydrocephalus. Its pathophysiology is still poorly understood. Diagnosis could be made prenatally or postnatally using ultrasound, CT, or MRI. The specific and precise diagnosis of this pathology requires the understanding of the typical radiological signs, in the prenatal and postnatal periods. There is no cure for hydranencephaly, and the prognosis is poor.

Declarations of Interest

The authors declare that they have no conflicts of interest.

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Volume 6 Issue 12 December 2023