



WESTERN NIGERIA JOURNAL OF MEDICAL SCIENCES (WNIJMS)

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OF SEVERE HYDROCEPHALUS**

HYDRANENCEPHALY IN A 2 MONTHS OLD INFANT WITH A WORKING DIAGNOSIS OF SEVERE HYDROCEPHALUS

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ABSTRACT

Introduction: Hydranencephaly is a rare central nervous system disorder in which some portions of the cerebral hemisphere are destroyed and replaced with cerebrospinal fluid. It is a lethal condition most often caused by thrombosis of the cerebral arteries or less commonly an in-utero infection. Affected infants present with severe hydrocephalus with or without regions of cortical mantle if destruction is ongoing, making diagnosis a challenge.

Case Report: Hydranencephaly in a 2 months old infant with a working diagnosis of severe hydrocephalus is presented.

Conclusion: The case highlights the value of medical imaging in the evaluation, diagnosis and management of a challenging clinical condition.

Keywords: Hydranencephaly, Hydrocephalus, Cortical mantle

INTRODUCTION

Hydranencephaly is a rare and severe central nervous system disorder characterized by partial or complete absence of the cerebral hemispheres, with preservation of islands of residual tissue at the occipital and orbitofrontal regions, choroid plexus, meninges and calvarium.¹⁻³ The condition is due to occlusion of both internal carotid arteries, and occurs in utero between 8 and 12 weeks of gestation.^{1,4}

Hydranencephaly occurs in less than 1 per 10000 births worldwide,^{1, 2} and is seen in 0.2% of infant autopsies.⁵ Most children with hydranencephaly die before their first birthday, however a small number attain second to third decade of life,¹ with the longest living patient attaining 32 years of age.¹

Supratentorial brain structures supplied by the anterior and middle cerebral arteries are usually absent, whereas basal and infratentorial brain structures supplied by the posterior circulation are preserved and include basal ganglia, thalamus, brainstem and cerebellum.^{1, 2, 4, 6} The absent brain is replaced by cerebrospinal fluid with the development of hydrocephalus of varying severity.

The present case was being managed as a case of congenital hydrocephalus from an ultrasound diagnosis until it was discovered after computed tomography (CT) to be a case of hydranencephaly, a rare congenital encephalopathy.

CASE REPORT

D.A. is a 2 months old male infant that presented at the pediatric clinic with complains of increasing head circumference since birth. Parents said they noticed the progressive increase in the head size after the 7th day of life. He was taken to General Hospital, Shomolu, before he was referred to the Lagos state University Teaching Hospital. Patient was the first child of both parents. The

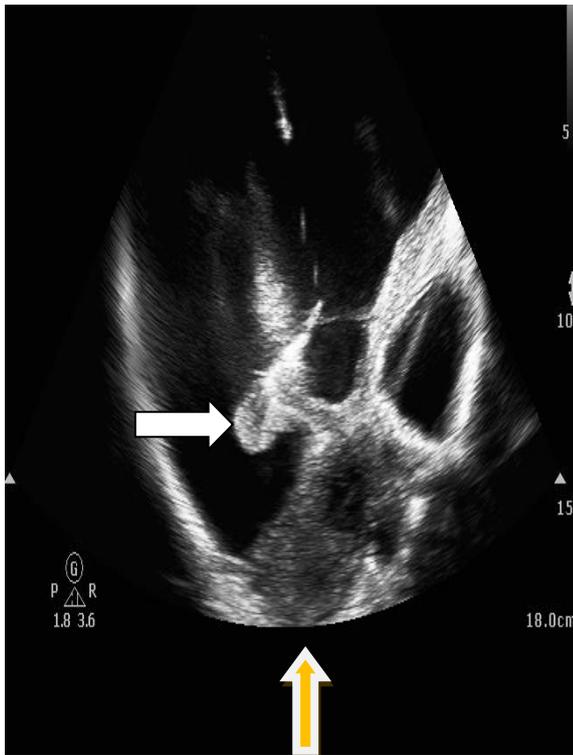


Figure 1: Coronal view (transfontanelle) ultrasound image showing absence of the cerebral mantle and replacement with cerebrospinal fluid. The third and fourth ventricles are dilated. Posterior brain structures (cerebellum) (purple arrow) appear normal. Right thalamus (white arrow)



Figure 2: Coronal view transfontanelle ultrasound image of the brain showing presence of the midline falx (red arrow)

mother is a 31-year-old housewife and his father a 52-yr-old artisan. The child was delivered at a mission house with a birth weight of 3.2kg. No history of a prenatal ultrasound scan was given. There was no history of febrile illness, smoking, ingestion of herbal mixtures and alcohol during pregnancy, while routine antenatal drugs were taken during pregnancy (paracetamol, folic acid and haematinics).

On physical examination, he was not pale, afebrile, anicteric, acyanosed, not dehydrated and there was no pedal oedema. There was hypertonia (increased muscle tone) of the limbs with irritable cry and weak sucking reflexes.

Head circumference 54cm (>95th percentile). Vesicular breath sounds were elicited on auscultation. Cardiovascular system was normal and the abdomen had no palpable organomegaly.

The full blood count was normal. Cranial ultrasound (USS) showed dilated ventricles with midline falx echo, absence of cortical mantle and presence of thalamus, brainstem and cerebellum in the posterior fossa (Figures 1 and 2).

Cranial computed tomography (CT) showed severe hydrocephalus (HU 12) with absence of brain tissue in the calvarium (Figure 3). Residual brain tissue was seen in the occiput adjacent the falx abutting the tentorium cerebelli (Figure 4). The posterior fossa was filled with cerebrospinal fluid and depressed brain tissues (Figure 5).



Figure 3: Axial CT scan at the level of the lateral ventricles showing cerebrospinal fluid filled brain cavity with presence of the midline falx. No brain tissue seen



Figure 4: Parasagittal CT scan image showing some residual brain tissue at the occiput (red arrow) with preservation of the meninges (white arrow).

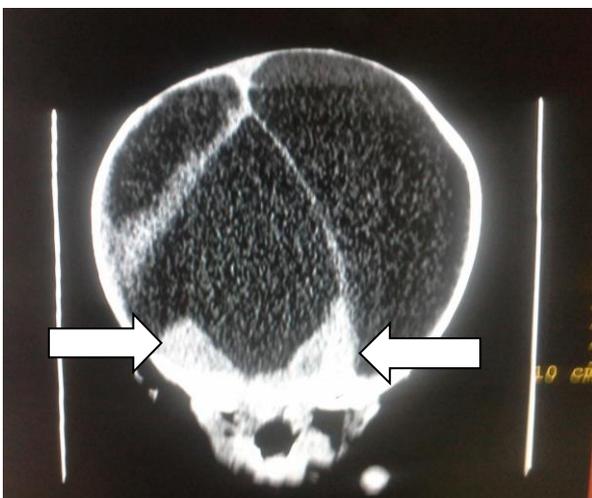


Figure 5: Coronal computed tomography image showing remnant posterior brain structures (white arrows)

DISCUSSION

Hydranencephaly was first described by Cruveihier in 1835. It develops secondary to occlusion of cerebral arteries above the supraclinoid level, mostly affecting the major vessels of the anterior circulation,⁷ resulting in absence and replacement of the cerebral hemispheres with cerebrospinal fluid (CSF) and necrotic debris.⁷ There was absence of the cerebral structures, with CSF filling the brain in the case presented.

Hydranencephaly can be bilateral or unilateral affecting one hemisphere (hemihydranencephaly).^{8, 9} Most cases are sporadic, involving 1 in 5000 pregnancies. It often occurs as an isolated defect without other associated malformations,⁷ as in the index case.

Several theories have been proposed for the etiopathogenesis of this neurological condition and they include the following:⁸⁻¹¹

1. Bilateral occlusion of the supraclinoid segment of the ICA or middle cerebral arteries
2. Extreme form of leucomalacia formed by confluence of multiple cystic cavities
3. Diffuse hypoxic-ischemic brain necrosis
4. Infection causing necrotizing vasculitis. Intrauterine infections, particularly toxoplasmosis and viral infections (enterovirus, adenovirus, cytomegalic, herpes simplex, Epstein bar and respiratory syncytial viruses. Toxic exposures and cocaine abuse have been reported.
5. Thromboplastic material from a deceased co-twin (monochorionic twin pregnancy).

The etiology in the index case could not be ascertained, but is likely due to the first theory postulated. Other causes have been attributed to ICA anomalies (aplasia and hypoplasia), genetic failures, drugs (estrogens and sodium valproate), young maternal age, deficiency of factor XII (fibrin stabilizing factor) and intracerebral hemorrhage. Cases with cerebellar hypoplasia and occlusion of bilateral posterior cerebral arteries have been reported¹². All reported causes could act directly or indirectly on the ICAs or in other cerebral districts causing profound brain alterations.^{2, 8}

Hydranencephaly is associated with various congenital anomalies including arthrogyriposis, renal aplastic dysplasia, poly-valvular heart defect, trisomy 13 and Fowler syndrome.^{5, 12} No other congenital anomalies were observed in the present case.

The infant with hydranencephaly usually appears normal at birth with intact brainstem reflexes such as sucking, swallowing, crying and movement of arms and legs, however a few weeks to months after birth, irritability, increased muscle tone, seizures, hydrocephalus, myoclonus and respiratory problems develop.^{10,13} The case presented had normal muscle tone and sucking reflexes at birth, as was observed by his mother, but after the first month he developed stiffness of the limbs (mother's own words) with weak sucking reflexes. She also noticed high pitched cry with reduction in awareness of her presence by the infant.

The clinical features of hydranencephaly are diverse and variable depending on severity. These include a vegetative state, delayed developmental milestone, intellectual deficits, spastic diplegia, spastic quadriparesis (paralysis), hypertonia and hypotonia of limbs, extensor plantar reflex, large head (>50cm), intact falx cerebri, ruptured falx cerebri.^{4,8,10} visual impairment, hypotelorism, optic nerve hypoplasia, chorioretinal atrophy, pigment clumping, dysplastic retina and blindness. The index case had large head circumference (54cm), intact falx cerebri and hypertonia of the limbs.

Other features are sensorineural hearing impairment, deafness and psychomotor retardation.¹² Subtle signs include feeble crying, difficulty with feeding and widened fontanelles.⁸ Systemic conditions like diabetes insipidus, adrenal insufficiency, venous malformation and central temperature regulation disorders have also been documented,² however no associated cranio-facial dysmorphism or limb deformity have been reported in hydranencephaly.¹²

There is usually delay in diagnosis due to normal clinical status and facial configuration at birth.^{2,10} Hydranencephaly can be diagnosed using ultrasound (antenatal and postnatal), magnetic resonance imaging (MRI) (antenatal and postnatal), Computed Tomography (CT),¹⁴ and skull transillumination.^{5,12} The index case had apparently normal head size and intact brainstem reflexes at birth. Postnatal

ultrasound and computed tomography of the skull was used for diagnosis in the case presented (Figures 1-5)

The differential diagnosis includes extreme hydrocephalus, alobar holoprosencephaly and porencephaly. Hydranencephaly can be differentiated from extreme hydrocephalus by a thin rim of cortical mantle in the latter, whereas alobar holoprosencephaly is associated with absence of falx and frequently coexisting midline facial abnormalities.^{4,8} The index case was initially being managed as a case of hydrocephalus at presentation, however a CT scan done showed dilated ventricles and splaying of the thalamus, with no rim of cortical tissue (Figure 1).

Prognosis is extremely poor with severe neurologic dysfunction, seizures, myoclonus, respiratory failure and death.⁷

Treatment for hydranencephaly includes ventriculo-peritoneal shunting and endoscopic choroid plexus cauterization. Ventriculo-peritoneal shunting has been tried with limited success because of the frequent need for shunt revision or removal.^{5, 12} The index case was scheduled for review with the imaging findings and other investigations by the neurosurgeon.

CONCLUSION

The index case highlights the value of medical imaging in the evaluation, diagnosis and management of a challenging clinical condition, with emphasis on the treatment and survival outcomes. The infant was referred to the neurosurgical unit for definitive management and care.

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