HYDRANENCEPHALY : CASE REPORT AND LITERATURE REVIEW

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ABSTRACT

Hydranencephaly is the total or virtually total absence of the cerebral hemispheres, which are reduced to membranous sac of glia tissue, with no ependymal coating, in an intact skull. This is rare disorder. It is classified as a circulatory encephalopathy from many causes (vascular, parasitic, viral,toxic,estrogenic,...).

It appears to be readily diagnoses prenatal by ultrasound. The neurological findings may be normal at birth. Transfrontanellar ultrasound,CT scanning and anatomical confirmation can established the diagnosis. The prognosis is hopeless and there is no treatment. Our report presents one case of hydranencephaly, clinical presentation and differential diagnosis from other common congenital diseases.

INTRODUCTION

"Hydranencephaly" is a term derived by the combination of two words, " hydrocephalus" and "anencephaly".[1] While hydrocephalic pathology is undoubtedly present in some cases, "anencephaly" is not quite appropriate, since the cranial vault and meninges are intact.[2-3] Hydranencephaly is a rare, isolated abnormality occuring in less than 1 per 10,000 births worldwide. It is the most severe form of bilateral cerebral cortical destruction. The exact etiology of hydranencephaly is unclear. The most popular etiological mechanism is intrauterine infarction of cerebral structures, primarily due to occlusion of the supraclinoid internal carotid artery.[4-7] The hydranencephalic infant may look remarkable normal at birth and demonstrated few neurologic abnormalities other than the lack of fixing and following an object with the eyes. CT scanning or cranial ultrasound examination should make the definitive diagnosis. CT offers a unique diagnostic tool for the evaluation of these infants. Most of the affected infants die within one year of life, but survival past three years has been reported.[1-3]

CASE PRESENTATION

This female infant was the first child of a 34 years old woman. Pregnancy was uncomplicated during antenatal care to term pregnancy about 39 weeks gestation. During delivery, poor maternal effort, so vacuum extraction (V/E) was performed. Cesarean section (C/S) was the final treatment due to fail V/E. At birth, Apgar score at one and five-minutes were nine and ten respectively, active child and normal in appearance. Her birth wedge and length were appropriate for gestational age. Two days after delivery, an abnormally slightly increasing head circumference was observed. Cranial ultrasound showed massive ventricular dilatation. A non-contrast enhanced CT scan demonstrated disruption of falx cerebri, absence of cerebral hemispheres, which were replaced by a large fluid-filled cavity, sparing normal thalamus and posterior fossa including the cerebellum, consistent with diagnosis of hydranencephaly.

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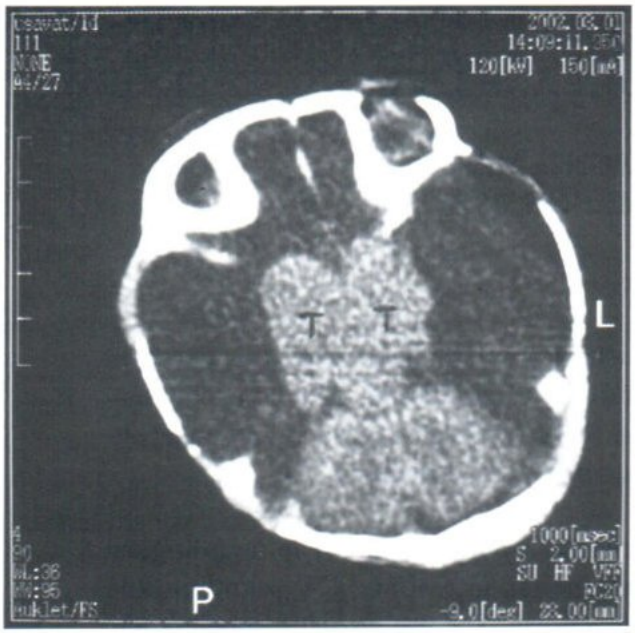
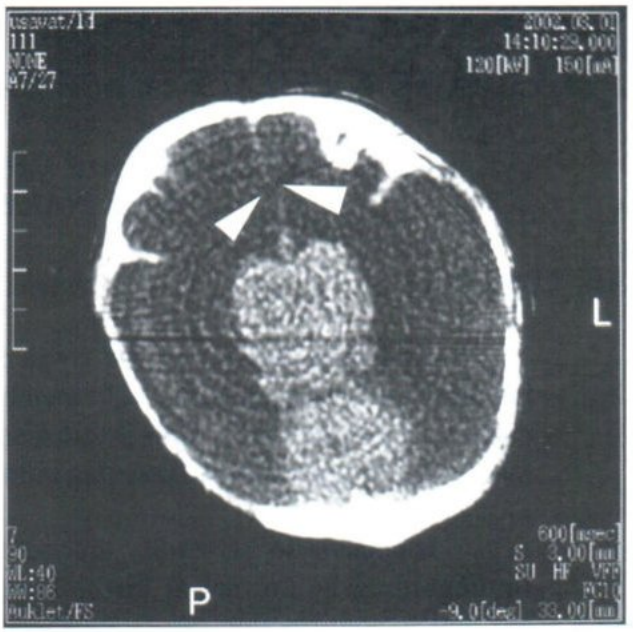
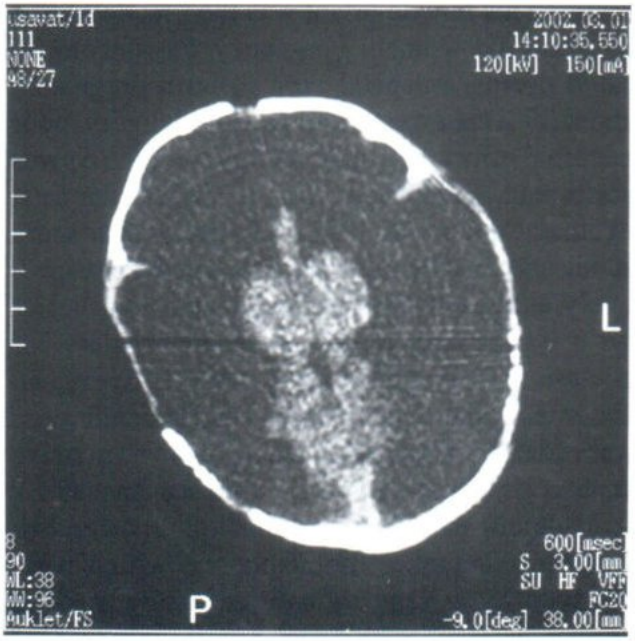


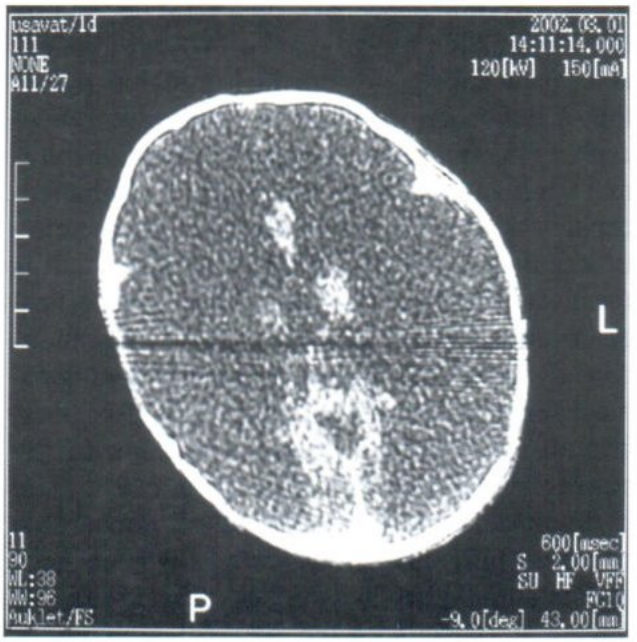
Slide 1 : CT scan of this newborn's head without use of Intravenous contrast axial slide 1 Transaxial view through the base shows normal thalami (T) and normal posterior fossa, including cerebellum.



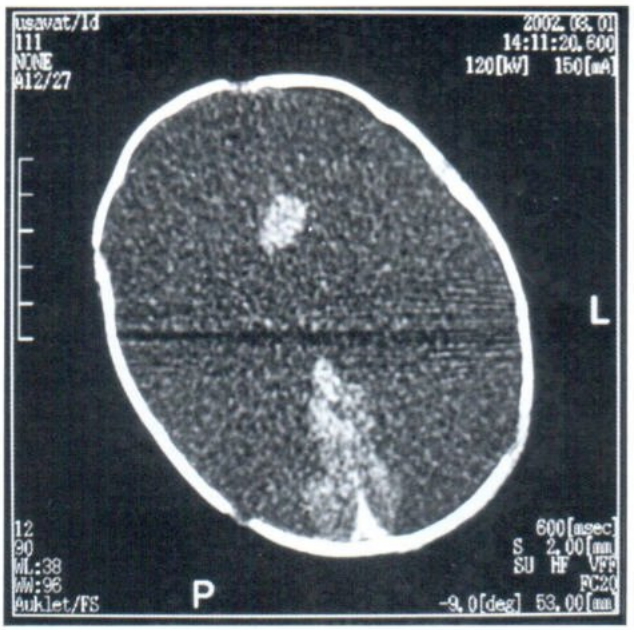
Slide 2 : Disrupted anterior falx is present (Arrow) Preservation of the falx cerebri helping to differentiate this from holoprosencephaly.



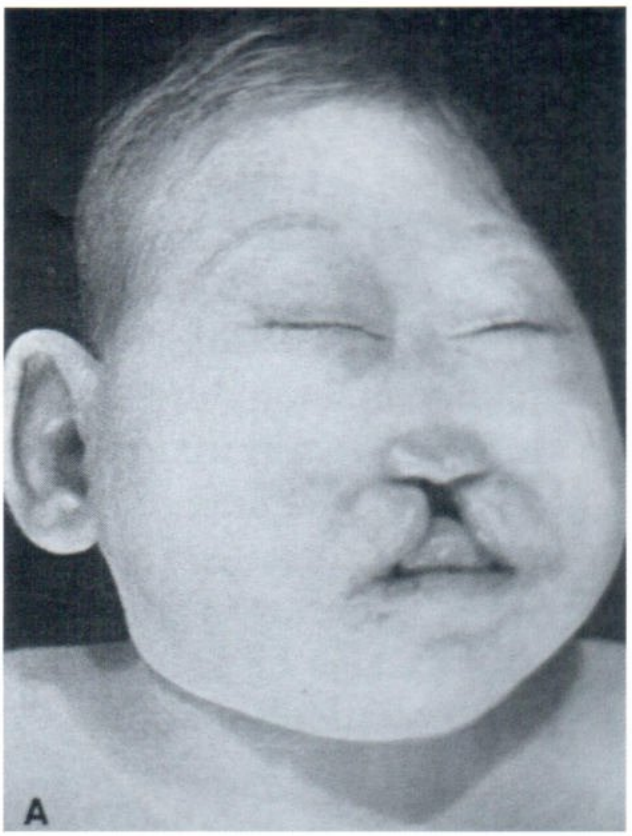
Slide 3 : A centrally placed tissue structure resembling a "brain stem model" represents diencephalic (thalamic and hypothalamic) and upper cerebellar and tentorial structures.



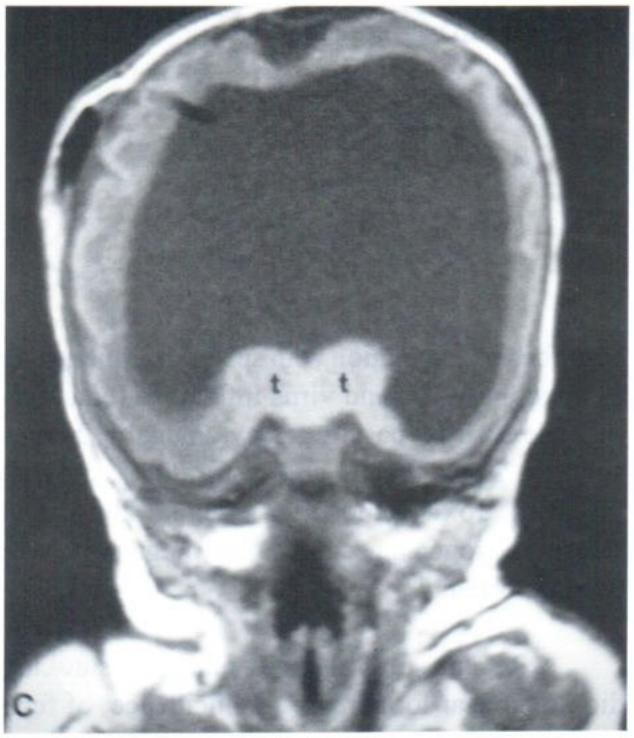
Slide 4 : A slightly higher levels show the falx intact posteriorly.



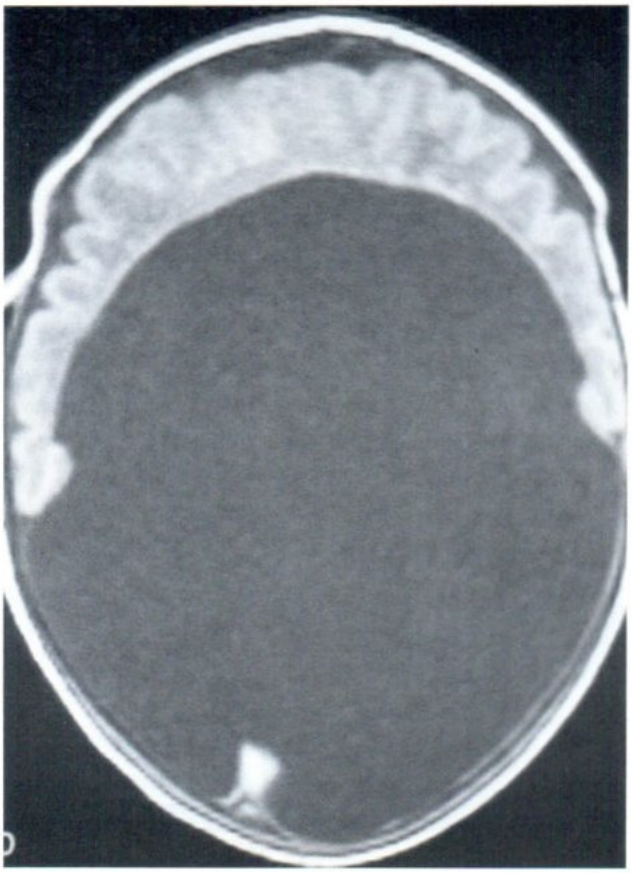
Slide 5 : This slide showing some occipital lobe preservation on either side of the posterior falx cerebri.



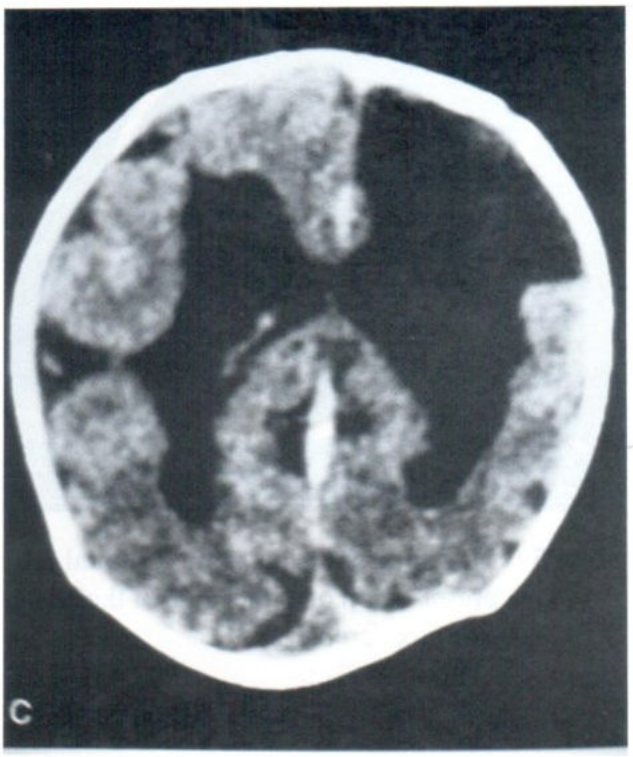
Slide 6 : Photograph of stillborn child with alobar holoprosencephaly shows low midline facial cleft, hypotelorism, and microcephaly. (Figure 6-9 from Diagnostic Pediatric Neuroradiology,1996)



Slide 7 : Coronal MRI T1–weighted image showing horseshoe – shaped monoventricle with fused thalami (t). The corpus callosum is absent and there is con tinuation of gray and white matter across the midline. The interhemispheric fissure is rudimentary and the falx is absent.



Slide 8 : Axial MR T1–weighted image in a different patient shows monoventicle and majority of residual cerebral tissues to be located rostrally. The interhemispheric fissure and falx are absent. There is a large dorsal midline cyst that is contiguous with the venticle



Slide 9 : Axial noncontrast computed tomography shows bilateral (left > right) open - lip schizencephalies.

DISCUSSION

Hydranencephaly is defined as the congenital absence of the cerebral hemispheres which are replaced by a large fluid - filled cavity but with intact cranial vault and meninges. A specific, clinical, nonradiographic diagnosis is usually impossible. The pathogenesis of hydranencephaly is thought to be a vascular accident, this cannot always be confirmed because internal carotid arteries are not always occluded at autopsy.[2] Massive cerebral destruction has been described in association with specific entities, such as, toxoplasmosis and virus infections (enterovirus, adenovirus, parvovirus,cytomegalic, herpes simplex, Epstein-Barr and respiratory syncytial visuses).[8] Toxic exposures, cocaine abuse and massive estrogen ingestion during pregnancy have been reported.[8-10]

Recent study[11] found maternal smoking did not appear to increase the incidence of fetal

congenital CNS anormalies overall, but might be associated with particular vascular patterns of damage to the developing brain that could predispose to a hydranencephalic malformtion. In monochorionic twin pregnancies, death of one twin in the second trimester may cause a vascular exchange to the living twin through the placental circulation, leading to hydranencephaly in the surviving fetus.[12]

Detection of hydranencephaly and other neural tube defects before birth has been enhanced by the development of a new screening technique for assaying the amniotic fluid. However, this test is not universally applied at present, and it is only accurate from the 16 th to the 22 nd week of pregnancy, a period dangerously close to the maximum allowable gestational age for legal abortion.[13-15] Thus, hydranencephalic fetuses continue to be carried to term, although there are no reliable statistics for the exact number. Table 1 summerized the clinical and radiographic finding hydra-nencephaly, severe bilateral porencephaly, and pictorially similar entities.

Clinically, hydranencephaly usually presents with normocephaly rather than macrocephaly, but occasionally may even be associated with microcephaly.[16-18] Transillumination of the head is nonspecific. The appearance of the child may be of some help in identifying infants with suspected hydranencephaly. Alobar holoprosencephaly always presents with midline cleft deformities such as fissured lips and/or palate, and hypotelorism.[17-18] A peculiar gyrated scalp was reported in one case of hydranencephaly but not with similar entities.[19] Previous recently reported one hydranencephalic infant associated with vascular malformation (port wine stains, generalized nevus flammeus, anormalous retinal vessels and absent internal carotid flow)[20] and one asscociated with dysplastic kidney and andramnios.[21]

Since hydranencephaly occurs after the brain and ventricle have fully performed, usually in the second trimester and the brain destruction is complete or almost complete in a bilateral internal carotid artery distribution with preservation of structure fed by posterior circulation. Plain radiograph findings of Hydranencephaly are usually nonspecific. Diagnostic ultrasound is useful in predicting the intrauterine process of macrocephaly, demonstrating abnormally increased echo-free areas of the intracranial contents, such as seen in hydranencephaly and hydrocephalus. Color Doppler ultrasound may shows absence of arterial flow above the supraclinoid portion of the internal carotid arteries, suggesting the diagnosis.[22]

CT and MRI help to accurate diagnosis and some authors suggested MRI is the modality of choice.[22] The cerebellum almost always intact, the brain stem usually atrophic, absence of cortical mantle, the thalamic, hypothalamic and mesencephalic structures are usually preserved and project into the cystic cavity. The choroid plexus,falx cerebri and tentorial cerebelli are usually intact but the falx cerebri may be deviated. With most of the cerebral cortex absent. The fetal head would be expected to be small. Although this may occur, the head is more often normal or increased in size because the choroid plexuses within the lateral ventricles continue to produce cerebral spinal fluid that is not adequately absorbed. This causes increased pressure, which may expand the head and lead to rupture of the falx cerebri.[23] Both of these findings were present in this case.

Angiographically, hydranencephaly demonstrates hypoplasia of the supraclinoid internal carotid arteries, with a normal external carotid vasculature. These changes were first described by Thelander et al in 1953 and later confirmed by others.[24-28] Remnants of subfrontal cortex may be present, supplied by a tangle of vessels in the anterior cerebral distribution. More

frequently, basal portions of the occipital or temporal lobes are present, along with relative normal hypothalamic and thalamic structures. There relative intact areas are supplied by normal posterior cerebral arteries. The cerebellum and vertebral-basilar arterial system are also intact.[7,16]

Pneumoencepahalography is rarely performed in hydranencephalic infants because of the danger of hydrocephalic mass effect and tonsillar or brain stem herniation. Ventriculography reveals no appreciable cerebral cortex, except for occasional remnants of occipital, temporal or subfrontal tissue.

An abnormal visual evoke response (VER) has been suggested in one case. It is not clear whether this infant was truly hydranencephalic. Two other patients had normal VER.[29] EEG measurements usually show diffusely abnormal patterns.[17,30]

Hydranencephaly may, on first impression, mimic severe hydrocephalus(dilated lateral ventricles).[3] Depending on the level of obstruction, concomitant dilatation of the third and fourth ventricles may be seen. The incidence of hydrocephalus approaches 1 in 1,000 births. Althrough there are many causes, the most common is an Arnold-Chiari typeII malformation secondary to a spina bifida. The most severe cases, however, are usually secondary to aqueductal stenosis. Hydrocephalus is often not isolated anormaly and can be associated with other intracranial abnormaities ,multiple anormaly syndromes, and abnormal karyotype.[31] With hydrocephalus, as with hydranencephaly, the head is normal to enlarged with an identifiable falx cerebri, which may be disruped in severe cases. Unlike in hydranencephaly, an intact rim of cortex is always present even in the most severe forms of hydrocephalus. It may, however, be difficult to identify prenatally. In aqueductal stenosis, a dilated third ventricle can often also be identified.[32]

Holoprosencephaly is developmental anormaly resulting from absent or incomplete diverticulation of the forebrain and occurs in 1 in 16,000 live births worldwide.[33] Holoprosencephaly is associated with a spectrum of facial anormalies and with visceral abnormalities with 75% of cases. The most severe form, alobar type, shows monoventricle communicates with a large dorsal cyst, no seperation of the ventricles, an absent falx, and fusion of the thalami. The convolutional markings are sparse and there is a smooth appearance to the brain surface. Migrational anormalies of the brain are often present. The cerebellum and brain stem are often spared. If the doppler is applied, absent anterior cerebral arteries or the occurrence of only a single vessel may be noted. There is absence of the internal cerebral veins, superior sinus, sagittal sinus and straight sinus.[34]

"Porencephaly", literally meaning" a hole in the brain," initially referred to cystic changes secondary to a vascular etiology.[17,23,35-36] However, the meaning of this word has been broadened to include any such changes from whatever cause. The porencephalic cyst is a focal area of cortical destruction.[2-3] When caused by middle cerebral artery infarctions, porencephalic cysts appear as bilateral fluid-filled clefts that communicate with the ventricles and is called schizencephaly. Unlike in hydranencepahly, both the frontal and parieto-occipital cortex are preserved. The falx cerebri is also preserved. The fetal head can be either normal or enlarged.

Other disesases such as massive congenital subdural hygromas and post-anoxic /infective encephalopathy may look like hydranencephaly as seen in table 1.

There are important reasons to differentiate hydranencephaly from hydrocephalus; these reasons related to prognosis and management.[35] Children with hydrocephalus, without chromo-

somal or other structural abnormalities, have an unpredictable prognosis. With proper ventricular shunt after birth, regrowth of the cortical mantle is usually evident of follow up imaging studies.[22] In contradistinction, hydranencephaly has an irretrievably poor prognosis, with only brain stem function remaining. Although most hydranencephalic children survive birth, they often die soon after. Rarely, these children may linger into their teenage years. If hydranencephaly were definitely diagnosed in utero, previous recently article[31] suggested cephalocentesis to decompress the fetal head, thus allowing a vaginal delivery to avoid the cesarean section, which thought to be unnecessary procedure, like in our case. On the other hand, if hydrocephalus were present, particularly without the presence of other anomalies, a cesarean section must be seriously considered.

SUMMERY

Hydranencephaly is rare disorder with poor prognosis and no treatment. The diagnostic ultrasound is helpful in screening intrauterine abnormality and children with abnormal cranial indices but non specific. The physical examination cannot suggest the disease due to the infant usually has normal face and less associated with other abnormalities. Nowadays CT or MRI are recommended for more precise differentiation and accurate diagnosis, angiography and other invasive procedures are unnecessary.

TABLE 1 : SUMMARY AND RADIOGRAPHIC FINDINGS IN HYDRANENCEPHALY AND PICTORIALLY SIMILAR ENTITIES

	Hydranencephaly (Classic)	Severe Hydrocephalus	Alobar Holoprosencephaly	Bilateral severe Porencephaly	Post- Anoxic/Infective Encephalopathy	Massive Congenital Subdural Hygroma
Clinical						
Cranial size at birth Or at discovery	Usually normal	Usually increased	Normal or microcephalic	Usually normal	Normal, micro-, or macrocephalic	Increased
Facial appearance	Normal, 1 case reported With gyrated scalp	Normal or with exophthalmos	Cleft palate defects, hypotelorism	Normal	Normal or "post viral"	Normal
Head Transillumination	4 +	4 +	3-4 +	3-4 +	1-4 +, depending on severity	4 +
Radiographic						
Plain skull Radiograph	Usually normal, occasionally macrocrania With open sutures	Sutures split. Associated abnormalities with Amold - Chiari syndrome (Lukenschadel)	Cleft defects, hypotelorism Normal or microcrania	Same as for hydranencephaly	With active disease, generalized cerebal swelling may split sutures. Inactive disease may present with split sutures (hydrocephalus) Or collapsed or overlapping sufures (atrophy)	Sutures split
Ultasonogram	Intact or partially intact falx with Echo - free fluid surrounding The central plexus cerebral tissue					
CT scan	Intact posterior fossa. intact or Relatively intact meninges & falx. falx may be midline or deviated But Not thickened. Majority of cranial vault is of CSF density, with remnants of temporal, occipital, or subfrontal cortex.	Some elements of cortex Usually identifiable. Massive CSF collection with intact falx	A frontal or occipital clump of Tissue. No falx. Generally intact posterior fossa. Midline osseous cleft defects	Wedge-shaped "infarcts" Without overlying cortex. Some identifiable ventricular remnants. Intact or relatively intact posterior fossa & meninges	Intact or peripheral areas of focal infaction. CT sequential changes in density with identifiable falx , ventricular structures, & areas of cerebral tissue	Mimics hydranencephaly. Wedge - shaped central collection of tissue
Pneumoencephalo-Gram and / or ventriculogram	Obstruction of the aqueduct or third ventricle	Variable		Same as for hydranencephaly		
Angiogram	Absence of supraclinoid Internal carotid arteries	Relatively intact cerebral vessels, massively compressed against the Cranial vault	Small, disordered tangle of vessels in frontal or occipital regions. Absence of falx vessels	Various degrees of midline or other focal arterial obstruction		Normal complement of supratentorial vessels compressed toward midline

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