Case Study

CASE REPORT: A RARE OCCURRENCE OF PREMATURE INFANT WITH HYDRANENCEPHALY IN INDONESIA

Azizah Hamida[1], Andi Arwan A[1]

Author's Affiliation:
1- Semen Gresik General Hospital, Indonesia

Correspondence:
Azizah Hamida, Email: azizahhamida@gmail.com

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ABSTRACT

Background: Hydranencephaly is a rare and mostly isolated central nervous system disorder characterized by the replacement of the cerebral hemispheres with cerebrospinal fluid. The etiopathogenesis is still unclear; with the occlusion of the internal carotid artery during fetal life is the most accepted hypothesis. This case occurs in less than 1 in 10.000 births worldwide.

Case Description: We are presented with a 1,300 g male infant who was born by induced labor at 30 weeks gestation due to fetal distress with prenatal and postnatal ultrasound findings led to the diagnosis of hydranencephaly which includes: replacement of cerebral hemispheres with fluid accumulation, the presence of disrupted choroid plexuses, falx cerebri, and no sign of cerebral mantles. Another subtle sign is the transilluminated head of the newborn.

Discussion: There is no clear reported data on the occurrence of hydranencephaly in Indonesia as most cases are terminated or expired before and during the delivery, which makes this is the first reported case of a premature infant survived vaginal delivery in Indonesia. The pathognomonic feature of hydranencephaly is the preservations of the posterior fossa structures. The preserved parts of the brain supplied by posterior cerebral and vertebral arteries support the hypothesis of internal carotid artery occlusion as the presumed etiology of hydranencephaly.

Conclusion: Early diagnosis of hydranencephaly with prenatal ultrasound is essential as it may help the family of the patient to process emotional issues and to distinguish hydranencephaly with other conditions related to prognosis and management.

Keywords: congenital anomaly, hydranencephaly, vascular disruption, premature

INTRODUCTION

For Systemic lupus Hydranencephaly is a rare, and mostly isolated central nervous system disorder characterized by complete or near-complete absence and replacement of the cerebral hemispheres by a sizeable amount of cerebrospinal fluid with the preservation of the skull and some parts of the brain.[1, 2]

The etiopathogenesis of hydranencephaly is still unclear; most research and literature support the hypothesis that the severe brain damage in hydranencephaly is related to early internal carotid artery involvement. It is thought caused by occlusion of bilateral internal carotid arteries in fetal life mainly during the second semester due to a variety of causes.[1–3] Hydranencephaly occurs in less than 1 in 5/10.000 births worldwide. There is no reported data on the occurrence of hydranencephaly in Indonesia.[1, 4]

CASE

A 20-year-old woman G2 P1 A1 L0 at 29 weeks of gestation with no symptoms and normal physical examination was admitted to precede further investigation following an altered obstetric ultrasound at her previous obstetrician. She had a history of uncomplicated spontaneous abortion at her first pregnancy. She denied having any previous disease with no family history of genetic or congenital anomalies. The patient did not take any unnecessary medicine during her pregnancy, did not have any history of previous smoking or any conditions that suggested congenital infections, drug abuse, or exposure to toxins. During this pregnancy, she

did not attend the local antenatal clinic regularly. She only did ultrasound once, and the obstetrician suspected hydranencephaly after ultrasound examination revealed abnormal intracranial anatomy of the fetus. The prenatal ultrasound at 29 weeks gestation revealed a sizeable amount of fluid replaced both of the fetus cerebral hemisphere, and the presence of disrupted choroid plexuses, third ventricle, and disrupted falx cerebri.[Figs. 1 and 2] The previous obstetrician presumed it as hydranencephaly with severe hydrocephalus as the differential diagnosis before referred the mother for further investigation at our hospital.

The mother delivered a male neonate by induced labor at 30 weeks gestation due to fetal distress. Apgar scores were 3/10, 5/10, and 10/10 at first, fifth, and tenth minutes, respectively. The birth weight was 1,300 g (appropriate for gestational age), head (occipito-frontal) circumference was 32 cm, and length of 48 cm. On examination, the newborn had an egg-shaped, transilluminated head[Fig. 3], which was macrocephaly or above 97th percentile on head circumference based on INTERGROWTH-21st charts. On the calvarium, there was a wide fontanel, which was soft and flat. Another deformity that we can found in the physical examination beside the head was the newborn's low set ears. The facial structure, neck, trunk, limbs, and genitalia appear normal without any obvious signs of deformity.

The baby was stable, pink in color, with no sign of respiratory distress. On the neurological examination, we observed that the neonate was lethargic. Both extremities flexed with very minimal resistance to the manipulation of the extremities. There was hypotonia in all the four extremities. In general, there was minimal response to a gentle touch and sound stimuli without any specific orientation. The Moro reflexes were incomplete and weak, with weak sucking and grasp reflexes. The baby crying was feeble and very weak, and he was rarely crying. The fundus examination of the neonate was normal with consistent pupillary reflex. The baby's eyes blink to the bright light stimuli. The milk was given to the baby from the feeding tube without any difficulty. The baby was stable and managed conservatively but passed away at 11 days old after a frequent apneic episode on his last day.

A postnatal ultrasound of the newborn's head performed one day after the delivery. The ultrasound revealed fluid collection in calvaria with the presence of disrupted falx cerebri, choroid plexuses, and residual cerebral parenchymal tissue at the occipital lobe, and there is no sign of cerebral mantle that distinguishes this condition with severe congenital hydrocephalus.[Fig. 4] The cerebellum is identified at the posterior fossae.[Fig. 5]

DISCUSSION

Hydranencephaly is a rare encephalosclastic anomaly characterized by the absence and replacement of the cerebral hemispheres with cerebrospinal fluid.[5] Hydranencephaly occurs in less than 1 in 10,000 births worldwide. There is no clear reported data on the occurrence of hydranencephaly in Indonesia as most cases are terminated or expired before delivery, which makes this is the first reported case of a premature infant outlived vaginal delivery in Indonesia.

The etiopathogenesis of hydranencephaly is heterogeneous, with the most accepted hypothesis is bilateral occlusions of the internal carotid arteries in the fetal life with evidence that the process might begin by as early as[8-12] weeks of gestation.[1,5] There is variable involvement on the inferior, frontal, temporal, and occipital lobes. The midbrain, cerebellum, thalami, basal ganglia, and choroid plexus are usually not involved. Falx cerebri usually present but may be disrupted or absent, with the septum pellucidum usually absent. Pathognomonic feature of hydranencephaly is the preservations of the brain stem and posterior fossa structures.[6] This finding explained that parts of the brain supplied by the posterior cerebral and vertebral arteries usually preserved, which supports the hypothesis of internal carotid artery occlusion mentioned above.[5, 6]

The diagnosis of hydranencephaly can be determined in utero by ultrasonography. Another gold standard modality for prenatal diagnosis is CT scan and MRI, which permit to differentiate more precisely hydranencephaly from holoprosencephaly or severe congenital hydrocephalus. In the absence of neuroradiological examination, transillumination could be useful for hydranencephaly diagnosis.[6–8] Early diagnosis of hydranencephaly with prenatal ultrasound is essential as it may help the family of the patient to

process emotional issues and to distinguish hydranencephaly with other conditions related to prognosis and management

It is crucial to differentiate between hydranencephaly with porencephaly, extreme hydrocephaly, and alobar holoprosencephaly. Hydranencephaly can be differentiated from extreme hydrocephalus by the presence of a thin rim of the cortical mantle around the cystic cavity, whereas alobar holoprosencephaly characterized by identifying the presence of falx and frequent coexisting midline facial abnormalities.[5, 6, 9]

Most reported case of hydranencephaly has a poor prognosis. Fetuses with hydranencephaly rarely survive to full term as most terminated before delivery. Most published case of hydranencephaly is delivered by elective caesarian section to avoid any obstetric complication from a possible cephalopelvic disproportion. Newborns with hydranencephaly can die at birth, but most infants die within the first year of their life. If the child survives, they will be severely handicapped.[10] This case is the first published case of prenatally diagnosed hydranencephaly, which survived normal vaginal delivery at a premature age in Indonesia.

CONCLUSION

In summary, we described a rare occurrence of premature infant survived vaginal delivery with prenatal and postnatal ultrasonography findings that described the diagnosis of hydranencephaly. The ultrasound examination also supports the hypothesis of internal carotid artery occlusion with the preservation part of the brain supplied by posterior cerebral and vertebral arteries. Early diagnosis of hydranencephaly with prenatal ultrasound is essential as it may help the family of the patient to process emotional issues and to distinguish hydranencephaly with other conditions such as severe hydrocephalus, porencephaly, and alobar holoprosencephaly related to prognosis and management.

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