Clinical Manifestations of Hydranencephaly: A Case in Monochorionic-Diamniotic Twin

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Hydranencephaly is a rare and fatal central nervous system (CNS) disorder, occurring in less than 1 per 10,000 births [1]. Cerebral hemispheres are almost absent and replaced with cerebrospinal fluid, but there is typically intact falx cerebri, thalamus, and cerebellum. Facial features and midline structures generally appear normal, distinguishing it from other CNS anomalies [1]. Most patients are stillborn or die within a few hours to months after birth, though long-term survival has been reported in rare cases [2]. Etiologies may be related to infections, toxins, genetics, or occlusive vascular lesion, but there are still many doubts about exact etiopathogenic mechanisms [1]. A previous case report briefly described the survival of a patient with hydranencephaly born as one of monochorionic-diamniotic (MCDA) twins [3]. However, there have been few reports on the neurological manifestations after survival especially as an extremely low birth weight (ELBW) infant of MCDA twins. Here, we report the clinical manifestations in a survived ELBW case of hydranencephaly, in MCDA twins.

The male patient was born at 33 weeks and 2 days of gestation. The birth weight was 840 g, height 33 cm, and head circumference (HC) 20.8 cm. His identical twin was born without any congenital anomalies and his birth weight was

1.7 kg (20th percentile). Prenatal ultrasound revealed the presence of fetal hydrops and brain ischemic insult. There was no maternal history of infections or exposure to toxins during pregnancy. Termination of pregnancy was avoided due to the possibility of intrauterine fetal death of the co-twin. The patient's Apgar score was 7 at 1 minute and 9 at 5 minutes. Physical examination at birth showed microcephaly, corneal opacities, anonychia of feet, but normal facial appearance (Fig. 1). He had stable spontaneous breathing and actively responded to external stimuli. Diagnostic tests were delayed due to his parents' initial refusal. As he was still alive with minimal care, further tests were performed. Brain ultrasonography performed at 18 days of age showed that supratentorial brain structures were almost absent, with the presence of only a small remnant of frontal lobe-like areas. At 28 days of age, 37⁺² weeks of postmenstrual age (PMA), he started showing some sucking movements, and echocardiography was normal. Glaucoma on both eyes was detected by eye consultation at 38 days of age. At 41⁺² weeks of PMA, a full dose of bottle feeding was possible, and automated auditory brainstem response test was normal. At 46⁺³ weeks of PMA, brain magnetic resonance imaging revealed the presence of the bilateral frontal lobes, small in size and displaced. A large cystic

Fig. 1. General appearance of the patient at 46 weeks of postmenstrual age. (A, B) He showed microcephaly, but normal facial appearance. (C, D) He had anonychia of feet.

Fig. 2. Brain magnetic resonance imaging of the patient. (A) T1-weighted sagittal image showed the cerebral hemispheres replaced by cerebrospinal fluid, but cerebellum and brainstem were within normal limits. (B) T2-weighted coronal and (C) axial image presented cystic area extending to the 4th ventricle with an asymmetric enlargement, with spared falx cerebri. (D) The lens in the left eyeball was not visualized on T2-weighted axial image.

area was observed in the brain, extending to the 4th ventricle with an asymmetric enlargement, with spared falx cerebri. The cerebellum and brainstem were within normal limits, and the lens in the left eyeball was not visualized (Fig. 2). Karyotype was a normal 46XY. On neurological examination, motor power showed grade 5 in all extremities, the muscle tone and deep tendon reflexes were normal. No pathologic reflexes were present. At 47⁺² weeks of PMA, he was discharged with a weight of 3.8 kg (< 3rd percentile), height 47.8 cm (< 3rd percentile), and HC of 31.5 cm (< 3rd percentile). At 6 months of age, on the last outpatient visit, he showed a weight of 6.7 kg (10th percentile), height 59.7 cm (< 3rd percentile), and HC of 37.5 cm (< 3rd percentile). He was still actively responded to external stimuli but showed fontanelle bulging and mild bilateral proptosis. He couldn't control his head and showed truncal hypotonia, but no spasticity of extremities.

In this case, we describe the clinical course of ELBW infant with hydranencephaly, in MCDA twins. Previous studies suggested that early severe bilateral internal carotid artery occlusion may induce

hydranencephaly [1,2]. However, MCDA twins have unique, different mechanisms, such as selective fetal growth restriction and twin-to-twin transfusion syndrome [4]. In an adverse position of the MCDA twin, exposure to hypoxia at some point in the fetal development process may lead to massive tissue necrosis with large cavitation. Rarely, patients with prolonged survival have been reported. Hence, documentation of neurologic conditions of survival in infants with hydranencephaly is essential for appropriate parental counseling on the possibility of prolonged survival and about the decision making of medical treatment.

Ventriculoperitoneal shunt or choroid plexus cauterization in infants can reduce brainstem compression and improve survival [2]. A retrospective study on 50 cases of hydranencephaly showed that the average life expectancy had increased due to appropriate medical and surgical treatments to 7.5 years in 58% of the subjects [1]. Bae et al. [5] reported the case of a patient with hydranencephaly who survived for 22 years and 6 months. They suggest that such patients can live beyond adolescence to adulthood. However, even

if surgery may prolong their lives, patients and their families face serious medical conditions due to recurrent pneumonia, seizures, intellectual disabilities, and spastic paralysis throughout the rest of their lives [1]. Counseling parents about poor prognosis and possible management options helps them to prepare for potential outcomes.

This study was approved by the Institutional Review Board of Kandong Sacred Heart Hospital (2020-12-004). Written informed consent by the patients was waived due to the retrospective nature of our study.

Conflicts of interest

No potential conflict of interest relevant to this article was reported.

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