HEMIHYDRANENCEPHALY OF PREMATURE INFANT - CASE REPORT

Tijana V Prodanovic[1,2] * and Suzana G Zivojinovic[1,2] *

[1]*University of Kragujevac, Faculty of Medical Sciences, Department of Pediatrics, Serbia*
[2]*Center for Neonatology, Pediatric Clinic, University Clinical Center Kragujevac, Serbia*

Received: 27.01.2023.
Accepted: 27.03.2023.

Corresponding author:

Tijana V Prodanovic

University of Kragujevac, Faculty of Medical Sciences, Department of Pediatrics, 69 Svetozara Markovica Street, 34000 Kragujevac, Serbia

Phone: +381 64 3990494

E-mail: tijanaprodanovic86@gmail.com

* Both authors contributed equally to this work.

DOI: 10.2478/sjecr-2023-0002

ABSTRACT

Hydranencephaly represents a rare anomaly of the central nervous system and is characterized by the parenchymal absence which is replaced with a membranous sac filled with cerebrospinal liquid, glial tissue and ependyma. When it is manifested as hemihydranencephaly, the patient's prognosis is better. We presented a case of a premature infant with hemihydranencephaly in twin pregnancy that had a fatal outcome. A premature infant in a controlled pregnancy was delivered at 31 weeks of gestational age. The mother of the newborn was hospitalized in the Clinic for Infective Diseases since she had tested positive for SARS-Covid-19. Following an urgent Caesarean section, the newborn was tested for Covid 19 using the rapid antigen and PCR test and the results were negative. In the delivery room, the tactile stimulation, aspiration, positive pressure ventilation of the infant were applied, after which it was intubated. After the intubation, a manual heart massage was performed, after which the cardiac activity and agonal breathing movements were detected." Three hours later, cardiorespiratory arrest happened, following unsuccessful resuscitation. The other twin was born with no anomalies, but showed signs of respiratory distress syndrome. Autopsy revealed that there was a thin-walled cavity filled with clear, yellowish liquid in the right hemisphere. Documenting cases of newborns with hydranencephaly is of a great importance for counseling parents regarding length of survival, successful management of pregnancy, and performance of prenatal diagnostic procedures.

Keywords: *Hydraencephaly, premature infant, twin pregnancy.*

INTRODUCTION

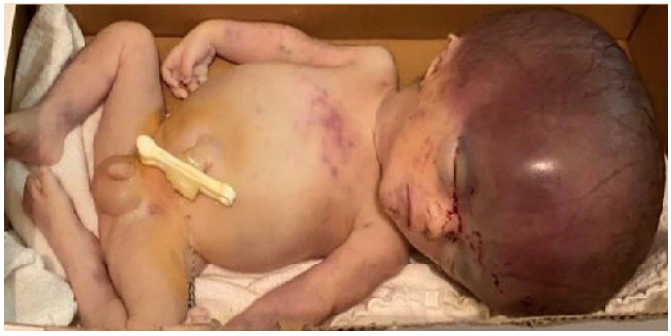
Hydranencephaly represents a rare anomaly of the central nervous system and is characterized by the absence of parenchyma that is replaced with a membranous sac filled with cerebral liquid, glial tissue and ependyma. The frequency of this anomaly is 1 in 10000 newborns (1, 2). It is commonly identified as bilateral schizencephaly, hydrocephalus, and alobar holoprosencephaly. Hydranencephaly occurs after the brain and ventricles are formed in the second trimester. Falx cerebri is present because the anomaly emerges after the formation of ventricles. Because of the preserved bloodstream in the hindbrain, thalamus, midbrain, basal ganglia and choroid plexus are preserved (3). Due to the parenchymal absence, it is expected that head size is small, however, it is increased because of the preserved choroid plexus and difficult reabsorption (4). Most common cause of the anomaly is carotid occlusion *in utero*. Teratogens that can attribute to the development of the anomaly include toxoplasmosis, viruses, cocaine abuse, and smoking (5,6). Hydranencephaly could manifests in the form of hemihydranencephaly - only one brain hemisphere is affected, hence the prognosis is better (7). This study shows an extremely rare clinical entity of a hemihydraencephaly with a fatal outcome. The aim of this study is to direct the attention to and to raise the awareness of this developmental disorder.

CASE STUDY

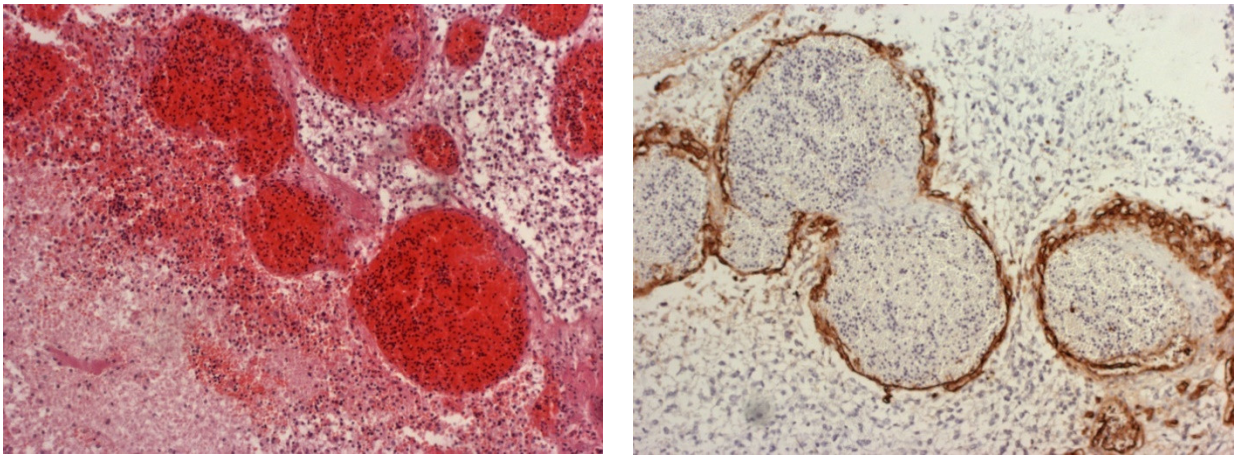
The seven month pregnant women was hospitalized in the Clinic for Infective Diseases because of the high temperature, respiratory problems, and positive test for SARS-Covid-19. She reported no chronic, systemic, and hereditary diseases, and denied the use of cigarettes and narcotics. Shortly before the delivery, ultrasound results showed signs of hydrocephalus, hydrops fetalis, and fetal bradycardia. Except for fetal bradycardia, the other twin did not show other deviations. Following a group discussion of clinicians, it was decided that the pregnancy should be ended by performing an urgent Caesarean section. A male infant was delivered (the first twin) at 31 weeks of gestational age (GA) via Caesarean section. His birth weight was 2750 g, the length 46 cm, head circumference 49 cm, chest circumference 32 cm, and Apgar score of 0/1 (0 at first, 1 at fifth minute). The newborn was unconscious, atonical, the skin was pale and cold with hematoma in the chest area and extremities, and it was not possible to cause primitive reflexes. The neck was short, the head discranic and macrocranic, soft to touch, and with increased occipital area, decreased size of the face with bent nasal root (Picture 1). Oral cavity and nostrils were unobstructed and filled with little blood content. In the abdominal area there were no signs of ascites were present. Examination showed no respiratory movements of the abdomen. Using the auscultatory method, no auditory breathing or heartbeats were detected with minimal values of oxygen saturation (SaO_2). In the delivery room, tactile stimulation, aspiration, positive pressure ventilation of the infant was applied, after which it was intubated. After intubation, a manual heart massage was performed, after which cardiac activity and agonal breathing movements were detected. Following reanimation, the newborn was tested for Covid 19 using the rapid antigen and PCR test from the samples from rectal and oropharyngeal swab, however the results were negative. Three hours after the intubation, cardiorespiratory arrest happened, following unsuccessful resuscitation. The other twin was born with no anomalies, but showed signs of respiratory distress syndrome.

Autopsy revealed that, after opening the cranium, brain tissue was softened with regions of diffuse bleeding. In the right hemisphere, a thin-walled cavity filled with clear, yellowish liquid was found. Basal ganglia and choroid plexus were preserved with the dilation of the lateral ventricles. Macroscopic examination of the other organs did not show the presence of anomalies.

Following microscopic analysis of brain tissue and Hematoxylin and Eosin staining protocol, the following signs were found: hyphemia, fresh blood in occipital regions, ventricular system of the left hemisphere, passive meningeal hyperemia with the defect of the dura mater along the sagittal line (Picture 2a). Immunohistochemical staining with the application of CD34 antibodies in the region of occipital ridge revealed multiplied and dilatated blood vessels with perivascular bleeding (Picture 2b).



Picture 1. Clinical representation of neonate with hemihydranencephaly



a) b)

Picture 2. Microscopic findings
a) hyphemia and fresh blood in occipital regions and the ventricular system of the left cerebral hemisphere
b) presence of multiplied and dilatated blood vessels with perivascular bleeding

DISCUSSION

Hydranencephaly as a clinical entity dates from the first half of the 19th century when Cruveilhier reported two cases of anencephaly. In one of the cases, the cranium was absent and in the other, hydranencephaly was clinically manifested (8).

The most common cause of the onset of hydranencephaly represents bilateral occlusion of internal carotid artery, which clearly differentiates hydranencephaly from hydrocephalus. Hydrocephalus manifests itself by the aforementioned vascular structures being present but changed. In 1969 an experiment on a monkey fetus that had undergone ligation of both carotid arteries and jugular veins took place. Then the fetus was inserted into the womb until spontaneous delivery was initiated. The ligations lead to the emergence of massive parenchyma necrosis and hemorrhages (9). Due to the one-sided occlusion of the internal carotid artery, hemihydranencephaly is formed which has a better prognosis. Dias et al. discussed the case in their case study of hemihydranencephaly in a 21-year-old patient (10). Hassanein et al. also reported hemihydranencephaly in their case study. They presented their case of 27-month-old female patient with hemihydranencephaly by analyzing cerebral white matter tract integrity lesions by the use of Quantitative Fibre Tracking Analysis by Diffusion Tensor Imaging (DTI) (2). Jordan et al. described the case of hydranencephaly diagnosed using CT angiography that clearly revealed vascular abnormalities. Their study indicated the benefits of multi slice CT angiography in the diagnosis of hydranencephaly compared to magnetic resonance imaging (MRI) for which patients need to be motionless (11). Conversely, Pavone et al. state that both multi slice CT and MRI are the gold standard for diagnosing hydranencephaly, showing preference for the magnet resonance imaging (7).

Prenatal viral infection is a common cause of hydranencephaly formation. In 1982, Bambirra et al. described four cases of toxoplasmosis that lead to the fatal hydranencephaly in newborns (12). Moreover, besides toxoplasmosis, viremia (adenovirus, cytomegalovirus, herpes simplex virus, enterovirus) may cause anomaly formation (13). In our case study, the mother of the newborn tested positive for Covid 19 immediately before delivery. Hemihydranencephaly represents an anomaly of the central nervous system that develops in the third trimester of pregnancy. Therefore, there is a possible correlation between Sars Covid 19 infection of the mother and the hemihydranencephaly of the newborn.

Systemic diseases that cause blood coagulation disorder may lead to developmental disorders of the central nervous system. McAdams et al. described the formation of hydranencephaly in newborns caused by the exacerbated maternal systemic lupus erythematosus that had led to blood hypercoagulation (14).

Twin pregnancy represents one of the risk factors for the emergence of hydranencephaly. The reason for the development of this central nervous system anomaly is a high degree of exposure to hypoxia in the gestational development with the causal development of massive necrosis of the brain tissue and formation of large cerebral cavities. The other twin did not show any signs of anomaly. Kim et al. obtained similar results in their study in which medical conditions of the other twin were similar to the conditions of the other twin in our study. Furthermore, in their study, the other twin was healthy (15). On the other hand, in the study by Huff et al., one of the twins was macrocranial, however, the results of magnetic resonance scan of the endocranium of both twins showed presence of hydranencephaly. The macrocranic twin died three weeks after birth (16).

Diagnosing hydranencephaly proves to be difficult even for experienced clinicians (17). A study shows a newborn who was diagnosed with hydranencephaly in the ninth month of life with the signs of high pressure of the cerebrovascular fluid (the setting-sun phenomenon, cutis marmorata and fontanel tension) (18). There are many anomalies of the central nervous system that differentially and diagnostically resemble hydranencephaly (alobar holoprosencephaly, hydrocephalus). The main difference is that the aforementioned anomalies (hydrocephalus, alobar holoprosencephaly) have a cerebral cortex around their perimeter, which is very thin in the extreme hydrocephalus, so it is often hard to see it prenatally (19).

In everyday clinical practice, increased head circumference of the newborns is mistakenly diagnosed as hydrocephalus. It is considered that 1% of all newborn diagnoses of hydrocephalus are consequently corrected and confirmed to be hydranencephaly (20, 21). Folver syndrome characterized by proliferative vasculopathy may resemble hydranencephaly in utero, which was described by Kline-Fath et al. in their study (22). Apart from the aforementioned study, a correlation between Robert's syndrome and hydranencephaly was described by Ekong et al. in their case study in 1978, in which the presence of hydranencephaly was determined at autopsy (23). Coban et al. described an unusual correlation between Poland syndrome and hydranencephaly in their case study. The cause of Poland syndrome is not clear, however, it is considered to be a bloodstream disorder at the level of subclavian artery. Also, carotid artery disorder is concidered to correlate between Poland Syndrome and hydranencephaly (24).

The prognosis for newborns diagnosed with this anomaly of the central nervous system is poor. There is a slightly better prognosis for patients with hemihydranencephaly, whereas a bilateral hydranencephaly most commonly ends fatally in utero or upon delivery.

CONCLUSION

Documenting cases of newborns with hydranencephaly is of utmost importance for counseling parents regarding length of survival, successful management of pregnancy, and performance of prenatal diagnostic procedures. Informing parents about the severity of the patients' medical condition and the poor prognosis helps them understand potentially fatal outcome. Despite the fact that there is a better prognosis for patients with hemihydranencephaly, our case study had a fatal outcome due to the infant's premature delivery.

REFERENCES

1. Gentry, Millicent MD; Connell, Mary MD. Hydranencephaly. Ultrasound Quarterly 2013; 29(3):267-268.
2. Hassanein SM, Abbas YA, Monib AM, El Alfy MS. Hemihydranencephaly syndrome: case report and review. Dev Neurorehabil. 2011;14(5):323-9.
3. Cater SW, Boyd BK, Ghate S V. Abnormalities of the fetal central nervous system: Prenatal us diagnosis with postnatal correlation. Radiographics. 2020;40(5):1458-72.
4. Kurtz A. JP. Case 7: Hydranencephaly. Radiology. 1999;210(2):419-22.
5. Rais-Bahrami K, Naqvi M. Hydranencephaly and maternal cocaine use: a case report. Clin Pediatr (Phila). 1990 Dec;29(12):729-30.
6. To WWK, Tang MHY. The association between maternal smoking and fetal hydranencephaly. J Obstet Gynaecol Res. 1999;25(1):39-42.
7. Pavone P, Praticò AD, Vitaliti G, Ruggieri M, Rizzo R, Parano E, Pavone L, Pero G, Falsaperla R. Hydranencephaly: cerebral spinal fluid instead of cerebral mantles. Ital J Pediatr. 2014;40:79.
8. Cruveilhier J. Anatomie Pathologique Du Corps Humain. 1st Editio. J. B. Baillière 1829-42, Paris; 1829.
9. RE. M. Brain pathology following fetal vascular occlusion: an experimental study. Invest Ophthalmol. 1969; 8(1):41-50.
10. Dias LS, Shivashankara KN, Vivek G. Hemihydranencephaly: Rare disease with key to secrets of the brain. BMJ Case Reports 2011; doi:10.1136/bcr.12.2010. 3658
11. Jordan L, Raymond G, Lin D, Gailloud P. CT angiography in a newborn child with hydranencephaly. J Perinatol. 2004 Sep;24(9):565-7.
12. Bambirra E., Pittella J. RM. Toxoplasmosis and hydranencephaly. N Engl J Med. 1982;302(18):1112-3.
13. Sandoval JI, De Jesus O. Hydranencephaly. [Updated 2022 Jun 27]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK558991/
14. McAdams RM. Maternal systemic lupus erythematosus and hydranencephaly in a neonate: A case report. J Matern Neonatal Med. 2005 Oct;18(4): 279-81.
15. Kim BG, Lim KJ, Yi YY. Clinical manifestations of hydranencephaly: A case in monochorionic-diamniotic twin. Ann Child Neurol. 2021;29(2):105-7.
16. Huff N, Naik S. Hydranencephaly in Monochorionic-Diamniotic Twins. Pediatr Neurol. 2017;67:107-8.
17. Omoto T, Takahashi T, Fujimori K, Kin S. Prenatal diagnosis of fetal microhydranencephaly: a case report and literature review. BMC Pregnancy Childbirth. 2020;20(1):688.
18. Petridis AK, Krause-Titz UR, Doukas A, Mehdorn HM. Delayed diagnosis of hydranencephaly in a nine-month-old child. Clin Pract. 2011 Sep 29;1(3):e65.

19. Wijerathne BT, Rathnayake GK, Ranaraja SK. A rare variation of hydranencephaly: case report. F1000Research. 2012 Oct 3;1:22.
20. Quek YW, Su PH, Tsao TF, Chen JY, Ng YY, Hu JM, et al. Hydranencephaly Associated with Interruption of Bilateral Internal Carotid Arteries. Pediatr Neonatol. 2008;49(2):43-7.
21. Moro T, Allyn W, Pipe L, Transilluminator N. C L I N I C A L V I S TA S Transillumination of the neonatal skull : seeing the light. 2002;167(11):1271-2.
22. Kline-Fath BM, Merrow AC, Calvo-Garcia MA, Nagaraj UD, Saal HM. Fowler syndrome and fetal MRI findings: a genetic disorder mimicking hydranencephaly/hydrocephalus. Pediatr Radiol. 2018 Jul 1;48(7): 1032-4.
23. Ekong CE u., Rozdilsky B. Hydranencephaly in Association with Roberts Syndrome. Can J Neurol Sci / J Can des Sci Neurol. 1978;5(2):253-5.
24. Coban D, Gunes T, Akin MA, Akcakus M, Yikilmaz A. Poland anomaly and hydranencephaly: An unusual association. Vol. 152 A, American Journal of Medical Genetics, Part A. 2010. p. 3182-4.