Hydrocephalus and Dandy-Walker syndrome. Clinical case

Andrea Michelle Canul Hilerio [1*] Alejandro Lenin Villalobos-Rodríguez[2] Yadira Hernández-Fuentes[3] Corina Bibiano-Rodríguez[4] Martín de Jesús Novelo-Salazar[5] Arturo Alfaro-Palma[6] Guillermo Padrón-Arredondo[7]

1. Medical Student Gyn-Obs Service General Hospital Playa del Carmen. Universidad Autónoma del Estado de Quintana Roo
2. Gyn-Obs. Service General Hospital Playa del Carmen. Orcid: 0000-0001-8410-0209
3. Public Health Master. Investigation Department General Hospital Playa del Carmen.
4. Gyn-Obs Nurse. Investigation Department General Hospital Playa del Carmen.
5. Degree in Nursing. Investigation Department General Hospital Playa del Carmen.
6. Hospital Administration Master. Investigation Department General Hospital Playa del Carmen.
7. General Surgeon. Investigation Department General Hospital Playa del Carmen.

*Correspondence: Alejandro L. Villalobos-Rodríguez

Received: 29 Nov 2023; Accepted: 05 Dec 2023; Published: 15 Dec 2023

Citation: Alejandro L. Villalobos-Rodríguez. Hydrocephalus and Dandy-Walker syndrome. Clinical case. AJMCRR 2023; 2(12): 1-5.

ABSTRACT

Introduction. *Dandy-Walker syndrome (DWS) is a developmental abnormality characterized by dilated posterior fossa, cystic enlargement of the fourth ventricle, hypoplasia of cerebellar vermis, and its upward rotation. The affected person may suffer from psychomotor retardation, ataxia, apnea attacks, muscle weakness, occasional muscle spasm, seizures, nystagmus, and macrocephaly. However, half of the cases have average intelligence.*

Clinical Case. *Male newborn at 36 weeks gestation obtained by cesarean section, with a history in the current pregnancy of poor prenatal control. During the fifth month of pregnancy, an ultrasound established a diagnostic suspicion of massive ventriculomegaly. At the sixth month, thinning of the cerebral cortex compression of the cerebellum in the posterior fossa was detected, and a possible ruling out of chromosomopathy related to Dandy-Walker Syndrome was suggested.*

Discussion. *Neurologically, this disease causes multiple anomalies, such as Hemorrhagic Stroke in a Young Adult with Undiagnosed Asymptomatic Dandy–Walker Malformation (5). The embryonic devel-*

opment of the cerebellum and fourth ventricle causes the tentorium to be displaced upwards, and the posterior fossa enlarges. The association of hydrocephalus with DWS is due to blockage of normal cerebrospinal flow, leading to an excessive amount of fluid accumulating in and around the brain and causing an increase in intracranial pressure and head circumference, which ultimately causes neurological impairment.

Conclusions. *The prognosis of patients with this syndrome depends on the association with intra/extracranial malformations. Early diagnosis of central nervous system abnormalities is considered one of the first objectives of obstetric ultrasound.*

Keywords: Dandy-Walker syndrome; hydrocephalus; Prenatal diagnosis.

INTRODUCTION

Dandy-Walker syndrome (DWS) is a developmental abnormality characterized by dilated posterior fossa, cystic enlargement of the fourth ventricle, hypoplasia of cerebellar vermis, and its upward rotation. The affected person may suffer from psychomotor retardation, ataxia, apnea attacks, muscle weakness, occasional muscle spasm, seizures, nystagmus, and macrocephaly. However, half of the cases have average intelligence.

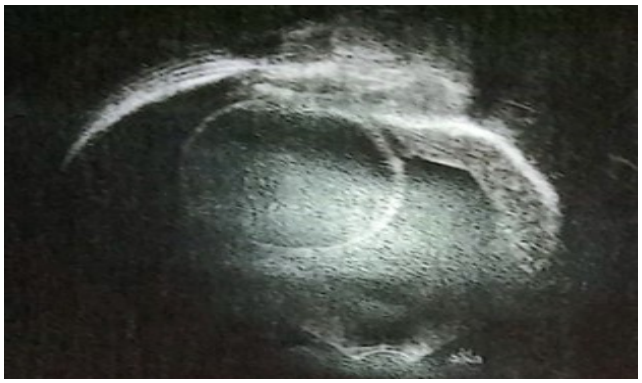
The etiology of DWS needs to be understood. However, the overlapping deletion of 3q24q25 has been reported in a limited number of patients. The frequency of DWS in the United States is approximately 1 per 30,000 live births, whereas the global occurrence is 6.79 per 100,000 childbirths. DWS may be asymptomatic or associated with diseases such as bipolar disorder, Acquired Immunodeficiency Syndrome, and kidney and liver diseases. Anesthetic management of DWS patients may face severe challenges due to the multi-organ association of craniofacial abnormalities, hydrocephalus, renal, and cardiac anomalies.

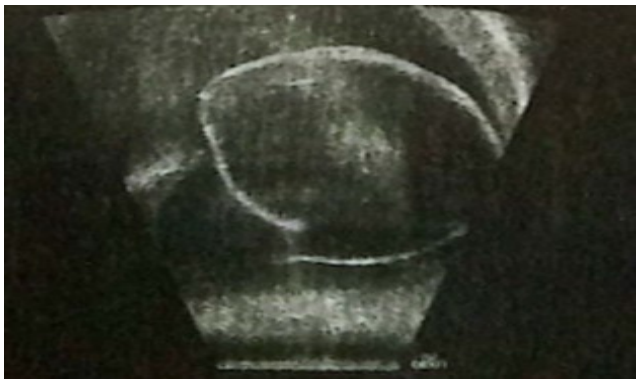
DWS or malformation is a congenital anomaly related to consanguinity, chromosomal abnormalities, infections, and alcohol consumption during pregnancy. The classic triad is presented with hypoplasia or agenesis of the cerebellar vermis, cystic dilation of the fourth ventricle, and hypertrophy of the posterior fossa with vertical displacement of the tentorium and transverse sinus. The prognosis depends on intra/extracranial malformations, being unfavorable for those who present these malformations, which are 50-70% of patients, with a mortality of 70% in fetuses, 14% in neonates, and 25% in infants. And favorable towards normal development within 50-65% of cases (1).

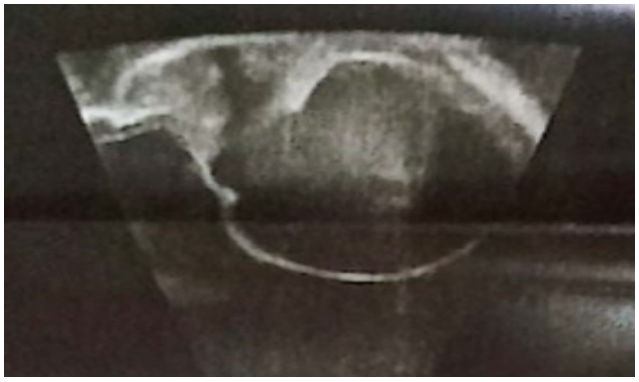
CLINICAL CASE

We have a male newborn at 36 weeks gestation obtained by cesarean section, with a history in the current pregnancy of poor prenatal control. During the fifth month of pregnancy, an ultrasound established a diagnostic suspicion of massive ventriculomegaly. In the sixth month, the cerebral cortex was thinning, cerebellum compression in the posterior fossa, and a possible ruling out of chromosomopathy related to Dandy-Walker Syndrome was detected. A transfontanellar and renal ultrasound was performed with the following findings: dilation of the cranial vault with anechoic fluid; almost total absence of cerebral hemispheres with little frantal

and temporal cortex with displacement of basal ganglia, midbrain and structures of the posterior fossa (Figs. 1-3).







The echocardiogram reports: perimembranous intraventricular communication of 1.9 x 1.4 mm with flow from left to right and systolic gradient of 6 mmHg.

DISCUSIÓN

During the past decade, fetal MR imaging has become a crucial part of the evaluation of the brain prenatally. It often plays a key role in counseling patients and postnatal management. Prenatal identification of a posterior fossa anomaly along the Dandy-Walker continuum does not have the same clinical implications as postnatally diagnosed classic DWS, making counseling and perinatal management challenging. Prenatally diagnosed isolated BP, in which the vermis is normal in size with an elevated tegmentovermian angle, generally portends a good prognosis; however, close follow-up is suggested because postnatal obstructive hydrocephalus can develop. Isolated vermian hypoplasia also can portend a good prognosis. However, prenatally diagnosed classic DWS can portend a worse prognosis, with an increased incidence of chromosomal and associated structural anomalies described [2].

Ultrasound is still the most effective method for prenatal diagnosis of DWS, which can be found during prenatal ultrasound screening in the second trimester. Previous studies believe that DWS should not be diagnosed prematurely because the cerebellar vermis develops from top to bottom at 17–18 weeks. It is considered to be a multifactorial genetic disease that is related to both genetic factors and environmental factors. There has been no significant sample size analysis of the chromosomal profile of DWS up to now [3].

DWS is described as a partial or complete agenesis of the cerebellar vermis, cystic dilatation of the fourth ventricle, and an expanded posterior fossa with an upward displacement of the lateral sinus, tentorium, and torcular herophili. Such a condition also confers diverse clinical features, from asymptomatic cases to severe intellectual disability. It might be related to other systems abnormalities in up to half of the individuals and may be associated with trisomy 18 [4].

Neurologically, this disease causes multiple anomalies, such as Hemorrhagic Stroke in a Young

Adult with Undiagnosed Asymptomatic Dandy–Walker Malformation [5]. The embryonic development of the cerebellum and fourth ventricle causes the tentorium to be displaced upwards, and the posterior fossa enlarges. The association of hydrocephalus with DWS is due to blockage of normal cerebrospinal flow, leading to an excessive amount of fluid accumulating in and around the brain and causing an increase in intracranial pressure and head circumference, which ultimately causes neurological impairment [6].

Neurocutaneous melanosis (NCM) is one of the rare phakomatoses. This childhood disorder has varied presentations and is associated with other Neurocutaneous syndromes like Sturge-Weber syndrome, Neurofibromatosis type 1, and DWS. The prognosis of NCM is dismal, especially when presented in adults and associated with DWS [7]. associated with ganglioglioma [8]; with Associated Neurofibromatosis [9]; Refractory psychiatric symptoms and seizure [10]; Hemorrhagic Stroke in a Young Adult [11]. Syringomyelia in an Adult [12].

Other sites of involvement include Bilateral Congenital Cataract [13], Ligneous conjunctivitis [14], buphthalmos, and vitreous hemorrhage [15]; in terms of associations with the musculoskeletal system, we have Scoliosis [16], cervical ribs [17]; of the digestive system: bilateral choanal atresia [18]; mental disorders [19, 20]; and others [21].

CONCLUSIONS

Anomalies of the central nervous system are considered one of the most common malformations seen on prenatal ultrasound. The prognosis of patients with this syndrome depends on the association with intra/extracranial malformations. Early diagnosis of central nervous system abnormalities is considered one of the first objectives of obstetric ultrasound. Hence, the importance of adequate prenatal control, due to its usefulness and accessibility, allows for assessing the adequate development of the fetus at the first level of care. Ultrasound signs of this pathology can be found in the first trimester of pregnancy.

Competing interests: All other authors declare that they have no competing interests.

Funding. None

REFERENCES

1. Oria MS, Rasib AR, Pirzad AF, Khel FWI, Khel MII, Wardak FR. A Rare Case of Dandy-Walker Syndrome. Int Med Case Rep J. 2022:15: 55-59.
2. Nagaraj UD, Kline-Fath BM, Horn PS, Venkatesan C. Evaluation of Posterior Fossa Biometric Measurements on Fetal MRI in the Evaluation of Dandy-Walker Continuum. Am J Neuroradiol. Sep 2021; 42:1716-21.
3. Sun Y, Wang T, Zhang N, Zhang P, Li Y. Clinical features and genetic analysis of Dandy Walker syndrome. BMC Pregnancy Childbirth. 2023; 23:40.
4. Ferreira de Souza LM, Galvão e Brito Medeiros A, Ribeiro Júnior JP, Nogueira de Melo A, Medeiros Dias SAM. Long Survival of a Patient with Trisomy 18 and Dandy-Walker Syndrome. Medicina. 2019; 55:352.
5. Abdul H, Burns J, Estevez A, Nasr El-Nimer C, Ekinde B, Lacaille S. Hemorrhagic Stroke in a Young Adult with Undiagnosed. Asymptomatic Dandy–Walker Malformation. Hindawi Case Rep Neurol Med. 2019; Article ID 1450703, 3 pages.

6. Jadhav SS, Dhok A, Mitra K, et al. Dandy-Walker Malformation with Hydrocephalus: Diagnosis and Its Treatment. Cureus. May 24, 2022; 14(5):e25287.
7. Sarwar M, Tripathy L, Jain H, Basu S, Sengupta A. Neurocutaneous melanosis with hydrocephalus and dandy-walker variant. Asian J Neurosurg. 2021; 16:876-80.
8. Wang LM, Zhang M, Wang PP, Zhou XG, Piao YS, Lu DH. A Dandy-Walker malformation associated with ganglioglioma. Chin Med J 2019; 132:2495-7.
9. Shah SP, Gohil JA, Kesavapisharady K, Easwer HV. Revisiting dandy–Walker malformation with associated neurofibromatosis. Asian J Neurosurg. 2021; 16:850-3.
10. Chen Y, Zhu J, Zhang D, Han L, Wang J, Yang W. Refractory psychiatric symptoms and seizure associated with Dandy-Walker syndrome: A case report and literature review. Medicine 2022; 101:46(e31421).
11. Zhang N, Qi Z, Zhang X, et al. Dandy–Walker syndrome associated with syringomyelia in an adult: a case report and literature review. J Int Med Res. 2019; 47(4):1771-7.
12. Charan GS, Narang GS, Kaur A, Kaur E. Dandy-Walker variant associated with bilateral congenital cataract. Int J App Basic Med Res. 2021; 11:277-9.
13. Srirampur A, Ramappa M, Chaurasia S, Vemuganti G. Ligneous conjunctivitis in a Dandy–Walker syndrome: A rare case report. Indian J Ophthalmol 2019; 67:143-5.
14. Qurban Q, Kamil Z, Saleem Tebha S, et al. Ocular manifestations in a patient with Dandy-Walker malformation: A case report. Radiol Case Rep. 2022; 17: 812-5.
15. Tsirikos AI, Wordie SJ. Dandy-Walker Syndrome Associated with Scoliosis: Clinical Presentation, Preoperative Assessment, and Treatment. Hindawi Case Rep Orthop. 2020; Article ID 8874819, 6 pages.
16. Schut P, Verdijk RM, Joosten M, et al. BMJ Case Rep 2018; 11:e225381.
17. Raghad K. Alsalamah, Mazyad M. Alenezi, Fahad Alsaab. Dandy-Walker syndrome with bilateral choanal atresia: A case report. Int J Surg Case Rep. 2022; 90:106702.
18. Porras Segovia A, Guerrero-Jiménez M, Carrillo de Albornoz Calahorro CM, et al. Psychosis and Dandy-Walker syndrome: a case report and review of the literature. General Psychiatry. 2021; 34:e100254.
19. Gama Marques J. Twenty years of misdiagnosis of schizophrenia in a patient with Dandy-Walker variant syndrome. General Psychiatry. 2019; 32:e100031.
20. Liu J, Liu Z, Yan H, Li Y. Dandy-Walker malformation in methylmalonic acidemia: a rare case report. BMC Pediatrics. 2021; 21:398.
21. Rajput R, Mishra S, Ahlawat P, et al. BMJ Case Rep. 2018; 11:e226281.