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Dandy Walker Malformation Prenatal Diagnosis and Postnatal Outcome in Multigravida: A rare Case

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ABSTRACT

Introduction: Dandy Walker malformation (DWM) is a rare congenital anomaly characterized by cerebellar vermis hypoplasia, posterior fossa expansion, and fourth ventricle enlargement, often associated with hydrocephalus and chromosomal abnormalities like Trisomy 18 (Edwards syndrome). This case report describes the prenatal diagnosis and postnatal outcome of DWM in a multigravida patient.

Case: A 43-year-old multigravida woman at 29–30 weeks' gestation presented to Dr. Zainoel Abidin General Hospital with suspected fetal anomalies. Obstetric examination revealed a transverse fundal height of 23 cm, estimated fetal weight of 1,500 grams, fetal heart rate of 140 beats/min, and maternal hypertension (160/90 mmHg). Ultrasound identified DWM (absent cerebellar vermis, enlarged fourth ventricle), bilateral hydronephrosis, and undescended testes. The patient had a history of poorly controlled hypertension and reported owning a cat for one year but denied alcohol or smoking. Following counseling, pregnancy termination was performed, resulting in the delivery of a 1,400-gram male infant (length: 36 cm, Apgar score: 4–5). Postnatal phenotypic examination revealed undescended testes, low-set ears, overlapping digits, respiratory distress, small stature, and hypotonia. Karyotyping confirmed Trisomy 18.

Conclusion: This case underscores the importance of prenatal ultrasound in detecting DWM and associated anomalies, enabling informed decision-making. The coexistence of DWM and Trisomy 18 highlights the need for genetic testing in such cases. Despite termination, the poor postnatal outcome reflects the severe prognosis of Trisomy 18. This report contributes to the limited literature on DWM in multigravida patients.

Keywords

Dandy Walker Malformation, Multigravida, Congenital Anomalies, Edwards Syndrome, Fetal Ultrasound, Postnatal Outcome

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INTRODUCTION

Dandy Walker Malformation (DWM) is a rare congenital anomaly affecting the posterior fossa structures of the brain, with an estimated incidence of 1 in 25,000–35,000 live births. It is characterized by a triad of features: hypoplasia or agenesis of the cerebellar vermis, cystic dilatation of the fourth ventricle, and enlargement of the posterior fossa, often accompanied by upward displacement of the tentorium cerebelli. These structural abnormalities disrupt normal brain development and cerebrospinal fluid (CSF) dynamics, frequently leading to hydrocephalus, ventriculomegaly, and increased intracranial pressure (ICP). DWM is often associated with additional congenital anomalies, including cardiac defects, renal abnormalities, genitourinary malformations, and craniofacial dysmorphisms, which complicate clinical management and prognosis [1-3].

The pathogenesis of DWM is linked to aberrant embryogenesis during the early stages of neural development, typically between the 6th and 8th weeks of pregnancy. The failure of the foramina of Luschka

and Magendie to open impairs CSF flow, resulting in cystic dilatation of the fourth ventricle and subsequent expansion of the posterior fossa. While the exact etiology remains multifactorial, genetic factors play a significant role, with DWM frequently associated with chromosomal abnormalities such as trisomy 18 (Edwards syndrome), trisomy 13, and other aneuploidies. Environmental factors, including maternal infections (e.g., cytomegalovirus or toxoplasmosis) and teratogenic exposures, have also been implicated in some cases, although the evidence remains limited [4-6].

DWM presents a diagnostic challenge because of its variable clinical manifestations, ranging from asymptomatic cases to severe neurological impairment. Prenatal diagnosis, primarily through fetal ultrasound and magnetic resonance imaging (MRI), is critical for identifying DWM and associated anomalies, enabling informed parental counseling and management. Postnatally, infants with DWM may exhibit developmental delays, motor dysfunction, seizures, and respiratory difficulties, particularly when complicated by hydrocephalus or chromosomal syndromes. The association with trisomy 18, a condition with a poor prognosis due to multiple organ system involvement, further underscores the need for comprehensive genetic evaluation in suspected DWM cases [7-10].

This case report describes the prenatal diagnosis and postnatal outcome of DWM in a multigravida patient, highlighting the diagnostic utility of fetal ultrasound and the complexities of managing DWM in the context of trisomy 18. By presenting detailed clinical, phenotypic, and genotypic findings, this report aims to contribute to the limited literature on DWM in multigravida pregnancies and emphasize the importance of early detection and multidisciplinary care in optimizing outcomes for such rare and complex conditions.

CASE REPORT

A 43-year-old G5P2 multigravida at 29–30 weeks' gestation was referred to the Obstetric Emergency Department at Dr. Zainoel Abidin General Hospital, Banda Aceh, for assessment of suspected fetal anomalies detected during routine prenatal care. On admission, the patient presented with elevated blood pressure (160/90 mmHg), pulse rate of 88 beats/min, and a respiratory rate of 20 breaths/min. Abdominal examination revealed a fundal height of 23 cm, estimated fetal weight of 1,500 g, breech presentation, and irregular uterine contractions. Speculum examination revealed a livid cervix with a closed external os and no evidence of vaginal bleeding or discharge. Vaginal examination confirmed a soft, posterior cervix with no dilatation and a floating fetal head in the uterus.

Fetal ultrasound revealed multiple congenital anomalies, including the absence of the cerebellar vermis, cystic dilatation of the fourth ventricle consistent with Dandy Walker malformation (DWM), bilateral hydronephrosis, and cryptorchidism. The patient had a history of chronic hypertension predating pregnancy and inconsistent adherence to antihypertensive therapy. She denied alcohol consumption or smoking but reported owning a pet cat for over one year. There was no history of recent infection or teratogenic exposure.

Given the severe fetal anomalies and maternal hypertension, multidisciplinary consultations involving obstetrics, neonatology, and genetic counseling were conducted. After an informed discussion with the patient and her family, the decision was made to proceed with pregnancy termination via cesarean section to mitigate maternal and fetal risks. A male neonate weighing 1,400 g and measuring 36 cm in length was delivered, with Apgar scores of 4 at 1 min and 5 at 5 minutes.

Postnatal examination revealed significant phenotypic abnormalities, including bilateral cryptorchidism, low-set ears, overlapping fingers, respiratory distress, generalized hypotonia, and short stature. Karyotyping confirmed trisomy 18 (Edwards syndrome), consistent with the observed congenital malformations and poor prognosis. The neonate was provided with supportive care in the neonatal intensive care unit, but further management details were not pursued because of the family's preference for palliative measures given the severity of the condition.

This case highlights the critical role of prenatal ultrasound in identifying complex congenital anomalies, such as DWM and associated chromosomal abnormalities. The coexistence of trisomy 18 underscores the importance of genetic testing in guiding clinical decision-making and counseling. The patient provided

informed consent for the use of clinical data for educational and research purposes, and ethical approval was obtained from the hospital's Institutional Review Board.

Figure 1. Dandy Walker Malformation Prenatal

DISCUSSION

DWM may occur as an isolated defect but is frequently associated with other congenital or chromosomal abnormalities, notably trisomy 18 (Edwards syndrome), as observed in this case. Trisomy 18, with an incidence of approximately 1 in 6,000 live births, is characterized by severe multisystem malformations, including craniofacial dysmorphisms (e.g., low-set ears), limb anomalies (e.g., overlapping fingers), cardiac defects, and genitourinary abnormalities (e.g., cryptorchidism) [11-13]. The neonate in this report exhibited these hallmark features, with postnatal karyotyping confirming trisomy 18, aligning with the poor prognosis typical of this syndrome, in which over 90% of affected infants succumb within the first year of life. The coexistence of DWM and Trisomy 18 underscores the importance of genetic testing in cases of complex congenital anomalies, as chromosomal abnormalities significantly influence clinical outcomes and management decisions [14].

Prenatal diagnosis of DWM relies primarily on obstetric ultrasound, typically performed in the second trimester, with sensitivity enhanced by advanced imaging modalities such as fetal magnetic resonance imaging (MRI). In this case, ultrasound at 29–30 weeks' gestation identified DWM and associated anomalies, including bilateral hydronephrosis and cryptorchidism, prompting comprehensive prenatal counseling. The late gestational age at diagnosis highlights the challenges in resource-limited settings, where access to early screening may be restricted. Maternal risk factors, such as advanced age (43 years in this case), are well documented for trisomy 18, with the risk increasing exponentially beyond 35 years. The patient's chronic hypertension, poorly controlled due to medication non-compliance, may have compounded obstetric risks, although its direct contribution to DWM or Trisomy 18 is unlikely. The reported cat ownership raised the possibility of zoonotic infections such as toxoplasmosis, a known teratogen associated with congenital brain anomalies. However, the absence of serologic testing or clinical evidence of infection limits causal inference, and this association remains speculative [15-18].

The management of DWM varies based on the severity of the anomalies, gestational age, and parental preferences. In isolated DWM cases with mild phenotypes, postnatal interventions, such as ventriculoperitoneal shunting for hydrocephalus, may improve outcomes. However, the presence of Trisomy 18, as in this case, shifts the focus to palliative care or pregnancy termination because of the lethal nature of the syndrome. The decision for cesarean section termination at 29–30 weeks was guided by grave fetal prognosis and maternal hypertension, which posed additional risks for continuing the pregnancy. Ethical considerations, including informed consent and family counseling, were critical in navigating this complex case and ensuring alignment with the patient's values and clinical realities [19].

This case highlights several clinical and research implications of the disease. First, it reinforces the pivotal role of prenatal ultrasound in detecting DWM and associated anomalies, thereby enabling timely decision-making. Second, it underscores the necessity of genetic testing to identify chromosomal abnormalities that can inform prognosis and management. Third, this case illustrates the challenges of managing rare congenital anomalies in multigravida patients, particularly in the context of advanced maternal age and comorbidities. The limitations of this report include the lack of fetal MRI to further characterize DWM and the absence of toxoplasmosis testing to explore environmental risk factors. Future research should focus on improving early prenatal screening, elucidating the genetic and environmental etiology of DWM, and developing standardized protocols for managing DWM with chromosomal syndromes. This case contributes to the sparse literature on DWM in multigravida pregnancies, advocating a multidisciplinary approach to optimize care in such complex scenarios [20].

CONCLUSION

Dandy Walker Malformation is a rare congenital brain anomaly that may be diagnosed prenatally using detailed ultrasound. It often coexists with other congenital defects and chromosomal abnormalities, such as trisomy 18. Early diagnosis allows for appropriate counseling, perinatal management, and decision-making regarding pregnancy continuation or termination.

DECLARATIONS

None

CONSENT FOR PUBLICATION

The Authors agree to be published in the Journal of the Society of Medicine.

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COMPETING INTERESTS

The authors declare no conflicts of interest in this case report.

AUTHORS' CONTRIBUTIONS

All authors contributed to the case report, including data collection, drafting, and review of the article. They approved the final version and were accountable for all the aspects.

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REFERENCE

1. Dandy WE, Blackfan KD. Internal hydrocephalus. An experimental, clinical, and pathological study. Arch Neurol Psychiatry. 1914;12(4):329–393.
2. Taggart JK, Walker AE. Congenital atresia of the foramina of Luschka and Magendie: A report of two cases with a survey of the literature. J Neuropathol Exp Neurol. 1942;1(2):89–105.
3. Klein O, Pierre-Kahn A, Boddaert N, Parisot D, Brunelle F. Dandy-Walker malformation: Prenatal diagnosis and prognosis. Childs Nerv Syst. 2003;19(7–8):484–489.
4. Moore KL, Persaud TVN, Torchia MG. The Developing Human: Clinically Oriented Embryology. 10th ed. Philadelphia: Elsevier; 2015.
5. Jones KL. Smith's Recognizable Patterns of Human Malformation. 7th ed. Philadelphia: Elsevier; 2013.
6. Ecker JL, Shipp TD, Bromley B, Benacerraf BR. The sonographic diagnosis of Dandy-Walker and Dandy-Walker variant: Associated findings and outcomes. Prenat Diagn. 2000;20(4):328–332.

7. Kolble N, Wisser J, Kurmanavicius J, Bolthauser E, Stallmach T, Huch A, et al. Dandy-Walker malformation: prenatal diagnosis and outcomes. Prenat Diagn. 2000;20(4):318–327.
8. Pascual JM, Solivera J, Prieto R, Barrios L, Lopez-Larrubia P, Cerdan S. Dandy-Walker malformation: Analysis of 19 cases. Neurologia. 2001;16(9):407–413.
9. Salihu HM, Kornosky JL, Alio AP, Druschel CM. Racial disparities in mortality among infants with Dandy-Walker syndrome. J Natl Med Assoc. 2009;101(6):550–556.
10. Hirsch JF, Pierre-Kahn A, Renier D, Sainte-Rose C, Hoppe-Hirsch E. The Dandy-Walker malformation. A review of 40 cases. J Neurosurg. 1984;61(3):515–522.
11. Osenbach RK, Menezes AH. Diagnosis and management of the Dandy-Walker malformation: 30 years of experience. Pediatr Neurosurg. 1992;18(4):179–189.
12. Estroff JA, Scott MR, Benacerraf BR. Dandy-Walker variant: Prenatal sonographic features and clinical outcome. Radiology. 1992;185(3):755–758.
13. Nyberg DA, Mahony BS, Kramer D, Forrest-Herman S, Fitzsimmons J. Prenatal diagnosis of the Dandy-Walker malformation and its variants: Sonographic and clinical correlations. J Ultrasound Med. 1988;7(8):421–428.
14. Springett A, Wellesley D, Greenlees R, Loane M, Addor MC, Arriola L, et al. Congenital anomalies associated with trisomy 18 in 12 European regions. Am J Med Genet A. 2015;167A(12):3062–3069.
15. Imataka G, Suzumura H, Arisaka O. Clinical features and associated anomalies in Dandy-Walker syndrome. Indian J Pediatr. 2007;74(5):519–522.
16. Lin MC, Chen SJ, Tsai TC, Tsai JD. Dandy-Walker syndrome: A review of 14 cases. Acta Paediatr Taiwan. 2000;41(4):192–196.
17. Bolduc ME, Limperopoulos C. Neurodevelopmental outcomes in children with cerebellar malformations: A systematic review. Dev Med Child Neurol. 2009;51(4):256–267.
18. Cuckle H, Wald N, Cuckle P, Platt LD. Maternal serum screening for trisomy 18 in the first trimester of pregnancy. Prenat Diagn. 1999;19(7):627–631.
19. Goetzinger KR, Cahill AG, Macones GA, Odibo AO. Association of congenital anomalies with trisomy 18: A retrospective cohort study. J Matern Fetal Neonatal Med. 2012;25(12):2701–2705.
20. Murray JC, Johnson JA, Bird TD. Dandy-Walker malformation: Etiologic heterogeneity and empiric recurrence risks. Clin Genet. 1985;28(4):272–283.