Significance of Timely Prenatal Care: A Case of Dandy-Walker Variant Diagnosed in the Third Trimester

Authors: German Andres Guevara Lizarazo, Jonattan Palacios Torres, Joshuart David Pascuas Varela , Mayra Alejandra Vargas Quiroga, Marbin Yulieth Alvarez Ramirez

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German Andres Guevara Lizarazo, Jonattan Palacios Torres[1], Joshuart David Pascuas Varela[2], Mayra Alejandra Vargas Quiroga[3], Marbin Yulieth Alvarez Ramirez[4]

andres1guevara@hotmail.com

Universidad Autónoma de Bucaramanga, Colombia.

Abstract

This case describes a 24-year-old primigravida woman from a rural area in Santander, Colombia, who began prenatal care late at 28.3 weeks. At 29.3 weeks, obstetric ultrasound showed hypoplasia of the cerebellar vermis and an enlarged cisterna magna, later confirmed by neurosonography and fetal magnetic resonance as a Dandy-Walker spectrum variant. Congenital infections were ruled out, and the mother declined invasive genetic testing. At 39 weeks, a cesarean section was performed, and the male newborn had adequate neonatal adaptation. Postnatal cranial ultrasound revealed a cystic dilation suggestive of a Dandy-Walker variant or Blake's pouch cyst. Outpatient follow-up was not possible. This case highlights how limited access to healthcare in rural areas can delay prenatal diagnosis and restrict a comprehensive approach, although some variants may have a benign neonatal course. Strengthening prenatal care access is essential for timely and adequate management.

Keywords: Dandy-Walker Syndrome; Hydrocephalus; Agenesis of Cerebellar Vermis; Holoprosencephaly; Fourth Ventricle.

[1] Universidad del Sinú, Colombia.
[2] Universidad de Magdalena, Colombia.
[3] Universidad del Sinú, Colombia.
[4] Universidad de Santander, Colombia.

Resumen

Este caso describe a una primigesta de 24 años, de zona rural de Santander, Colombia, quien inició el control prenatal tardíamente a las 28,3 semanas. A las 29,3 semanas, una ecografía obstétrica evidenció vermis cerebeloso hipoplásico y cisterna magna aumentada, confirmándose por neurosonografía y RM fetal como una variante del espectro Dandy-Walker. Se descartaron infecciones congénitas y la madre rechazó estudios genéticos invasivos. A las 39 semanas se realizó cesárea, naciendo un neonato con buena adaptación y hallazgo postnatal de dilatación quística posterior compatible con variante de Dandy-Walker o quiste de la bolsa de Blake. El seguimiento postnatal no fue posible. El caso resalta cómo el acceso limitado a servicios de salud en áreas rurales retrasa el diagnóstico prenatal y limita el abordaje integral, aunque algunas variantes de esta malformación pueden tener un curso neonatal benigno.

Palabras clave: Síndrome de Dandy-Walker; Hidrocefalia; Agenesia de Vermis Cerebeloso; Holoprosencefalia; Cuarto Ventrículo.

Introduction

Dandy-Walker Syndrome (DWS) is a congenital condition that affects brain structures, specifically the posterior fossa and cerebellum. It is characterized by imaging findings typically defined by a triad: partial or complete agenesis of the cerebellar vermis, enlargement of the posterior fossa, and cystic dilation of the fourth ventricle (Society for Maternal-Fetal Medicine & Monteagudo, 2020, p. B39). In the fetus, the partial or complete absence of the cerebellar vermis results in failure of the fourth ventricle to close, upward displacement of the vermis, and disruption in the development of the straight sinus, torculus, and tentorium. These alterations ultimately lead to posterior fossa enlargement and, consequently, hydrocephalus (Norton et al., 2020, pp. B30–B33).

The incidence of this malformation is approximately 1 in every 35,000 births. The most characteristic in utero imaging finding is hydrocephalus, but less than 10% of hydrocephalus cases are attributed to DWS (Society for Maternal-Fetal Medicine & Monteagudo, 2020, p. B39). This malformation occurs more frequently in female fetuses (Hart et al., 1972, pp. 771–780). Within the Dandy-Walker spectrum, other findings include holoprosencephaly, Blake's pouch cyst, ventriculomegaly, hypoplastic nasal bone, micrognathia, intrahepatic calcifications, hepatosplenomegaly, cardiomegaly, and perimembranous ventricular septal defect (Hart et al., 1972, pp. 771–780). In addition, it may also be accompanied by the presence of more common trisomies such as 9, 13, 18, and 21, and deletions of 6p and 3q22-q241. The presence of exposure to congenital infections within the TORCH spectrum is also documented in the literature, although at a very low incidence.

During embryogenesis, brain structures gradually take shape as the fetus develops. Therefore, a diagnosis of this malformation cannot be made before the 15th week of gestation, as it is estimated that the vermis completes its formation around that week. Obstetric ultrasound findings to define an enlarged cisterna magna are greater than 10 millimeters at the level of the transcerebellar plane (Norton et al., 2020, pp. B30–B33).

The range of imaging findings for this malformation can be diagnosed in utero by transabdominal obstetric ultrasound, in addition to supplementary imaging such as magnetic resonance imaging. After birth, the physical examination is reviewed for

the presence of microgantia, hypoplastic nasal bone, and heart murmur. This is complemented by transfontanellar ultrasound, brain magnetic resonance imaging, and monitoring by specialists such as pediatric neurology and neurosurgery.

Clinic Case

A 24-year-old female patient from a dispersed rural area of Santander, Colombia, with no significant medical, surgical or family history, and no consumption of psychoactive substances, consulted a general practitioner because she was late for her period. During this medical consultation, it was documented that the patient is pregnant with a G1P0 obstetric formula and her last period was due on 30/06/2024. The patient began her prenatal check-ups at 28.3 weeks of gestation due to her last period, and was not compliant with her prenatal check-ups because she lived in a dispersed rural area near the location where the consultations were held. During her first prenatal check-up on 01/22/2024, her gestational age was 29.3 weeks according to a second trimester ultrasound. Routine paraclinical tests were performed, which showed positive hemoclassification O, hemoglobin: 12.5 mg/dL, hematocrit: 37%, Leukocytes: 11,800 cells/m3, platelets 249,000, TORCH profile result: cytomegalovirus IgG: reactive, cytomegalovirus IgM: non-reactive, varicella zoster IgG: negative, treponema pallidum test: negative, IgG for trypanosoma cruzi: negative, HIV rapid test: negative, Hepatitis b HBsAg: non-reactive, rubella IgG: reactive, Rubella IgM: non-reactive, Toxoplasma gondii IgG: reactive, Toxoplasma gondii IgM: negative, fasting blood glucose 78 mg/dL, blood parasites: negative, vaginal discharge smear with no evidence of infection, urine culture: negative, thyroid profile normal. During that first consultation, the patient showed no abnormalities upon physical examination, vital signs within normal limits, for which an obstetric ultrasound was scheduled.

The patient had her first obstetric ultrasound on 01/22/25, which showed a live singleton fetus, cephalic presentation, longitudinal on the left dorsum, estimated fetal weight of 1365 grams, growing in the 30.8% percentile, amniotic fluid index of 15.2 cm, fetocardia 152 bpm, biometry for 29.3 weeks of gestation. Because the patient started prenatal checkups late, in addition to living in a dispersed rural area and not having adequate access to health care, she was indicated for priority evaluation by the maternal-fetal medicine service.

The patient was seen by the maternal-fetal medicine service on 01/22/25. A detailed anatomical ultrasound was performed, which showed the integrity of the skull, intracranial structures present: medical line, cavum, thalami, peduncles, cerebral ventricles and parenchyma of normal ultrasound appearance, 8 mm atrioventricular, normal posterior fossa, 12.7 mm cisterna magna, 5.9 mm nuchal fold, a decreased cerebellar vermis was evident. Male external genitalia. This study was concluded with a diagnosis of megacisterna magna, in addition to hypoplasia of the cerebellar vermis with no additional findings. The mother was informed of the findings and was offered the option of terminating the pregnancy, however she stated that she wishes to continue with her pregnancy. Studies to determine the extent of the malformation were offered by taking amniotic fluid samples. The patient stated that she does not wish to have amniotic fluid samples taken. Otherwise, she is advised to continue with periodic check-ups by maternal-fetal medicine.

New control by sub-specialty, neurosonography is performed, on 04/02/2025, when the gestational age was 32.3 weeks, a single cephalic fetus is evident, left dorsum, in the intracerebral evaluation, gyri and convolutions of lissencephalic appearance are observed, intact corpus callosum and adequate size, rectangular septum pellucidum cavity, normal lateral ventricles and adequate size, no calcifications or adhesions, normal cerebral peduncles, lateral ventricles within normal range, abnormal cerebellopontine angle in the posterior fossa, abnormal torcula, partial agenesis of the inferior cerebellar vermis with abnormal distribution of the cerebellar folia, enlarged cisterna magna. According to the above findings, the patient was indicated as a priority to perform magnetic resonance imaging of the fetal central nervous system.

The patient was sent to the emergency room on 22/02/2025 for a fetal brain magnetic resonance imaging. The results showed findings of cerebellar vermis hypoplasia, with an anteroposterior vermis diameter of 1 cm, a height of 1.2 cm, no alterations in the morphology of the cerebellar hemispheres, widening of the cisternum magna that communicates internally with the fourth ventricle, no dilation of the fourth ventricle: the conclusion of the study is: vermian hypoplasia compatible with Dandy-Walker variant.

The last visit by the subspecialist was on the 03/18/2025, where a new ultrasound control was performed: biometry: 37.3 weeks, estimated fetal weight: 3.131 grams growing in the 54% percentile, amniotic fluid index: 9.6 cm, normal fetal Doppler,

the patient was told that the pregnancy should be terminated around the 39th week of gestation. Because of this she was scheduled to go to the emergency room on 03/26/2025 for termination of the pregnancy by cesarean section.

The patient came to the emergency room on 03/26/2025, where a cesarean section was performed and the baby showed an Apgar score 8/10 at 1 minute of life and 10/10 at 5 minutes. As a result of gestation, the baby was hospitalized for further clinical studies. A transfontanelar ultrasound was performed with evidence of a transverse cerebellar diameter of 5.4 cm, a cystic dilatation located in the posterior aspect of the cerebellum measuring 3.8 x 1.6 cm, there was no compressive effect on the cerebellum or displacement. Conclusion of the study: findings at the cerebellar level suggestive of a Dandy-Walker variant vs. Blake's pouch cyst. During in-hospital surveillance, the patient with DWM always presented with physical examination findings of reactivity to stimuli, loud crying, vigorous sucking, mobilization of all 4 extremities with no evidence of abnormal movements, no seizures, sucking and rooting reflexes were present. No further information is available on the patient.

Discussion

The Dandy Walker malformation (DWM) spectrum is a very rare disease that occurs in 1 in 35,000 births and is characterized by the described imaging triad, which can be found from the 15th week of gestation (Zuluaga Jaramillo et al., 2009, pp. 147–153; Hart et al., 1972, pp. 771–780; Robinson et al., 2007, pp. 211–223; Bromley et al., 1994, pp. 761–763). In the present clinical case presented, it is described that the pregnant woman underwent ultrasound at week 29.3 of gestation due to late start of prenatal controls, in said obstetric ultrasound of anatomical detail, at the brain level, partial absence of the cerebellar vermis, cisterna magna of 12.7 millimeters (normal <10 millimeters 1), posterior fossa with abnormal cerebellopontine angle and abnormal torcula are evident, all of these findings characteristic of the aforementioned malformation corresponding to Dandy-Walker.

In this case it is also very important to mention that the product of the gestation is male, a fact that draws attention since it occurs in greater proportion in female fetuses (Zuluaga Jaramillo et al., 2009, pp. 147–153). The sex of the patient at birth was confirmed; he did not present any phenotypic characteristics related to

chromosomal abnormalities nor morphological abnormalities such as micrognathia or hypoplasia of the nasal bone during the inspection by pediatrics after birth.

In the case presented, the baby's mother did not allow additional diagnostic tests on amniotic fluid, such as FISH karyotype and chromosomal microarray (CMA) (Society for Maternal-Fetal Medicine & Monteagudo, 2020, p. B39). However, infectious causes were ruled out, since during the few prenatal checkups that were performed, the patient did not have an acute TORCH spectrum infection. In the previous case, the cause of this malformation could not be determined.

Since the diagnosis of DWM, the mother was offered serial obstetric ultrasounds to determine the progression of fetal growth and determine the presence or absence of worsening of the defect.

For live births diagnosed with DWM, transfontanellar ultrasound is performed in addition to imaging such as magnetic resonance. In this patient, a transfontanellar ultrasound revealed the presence of a cystic dilatation located in the posterior aspect of the cerebellum. The conclusion was that this was a Dandy-Walker malformation versus a Blake pouch cyst, which is a typical finding in the spectrum of posterior fossa malformations and DWM (Robinson et al., 2007, pp. 211–223).

In these patients, treatment is based on surgical ventriculoperitoneal shunting to reduce intracranial pressure. However, in this clinical case, the patient presented adequate neonatal adaptation at birth, with no signs of intracranial hypertension or hydrocephalus. At birth, the pediatrician determined that the patient presented adequate sucking, limb movements, respiratory and hemodynamic patterns. Subsequent follow-up was not possible because communication with the mother was not possible.

Conclusion

The late initiation of prenatal care can limit the timely and complete diagnosis of congenital malformations of the central nervous system. In this case, the patient began prenatal care at 28.3 weeks, which prevented early detection of the Dandy-Walker spectrum malformation and the performance of complementary studies such as genetic analysis (karyotype, FISH, or CMA) or amniocentesis, which

would have allowed for a more accurate etiological diagnosis and appropriate counseling for the pregnant woman.

The late identification of ultrasound findings suggestive of posterior fossa malformation reduces the opportunities for comprehensive fetal evaluation and informed decisions regarding pregnancy. The finding of an enlarged cisterna magna and hypoplastic cerebellar vermis at 29.3 weeks limited detailed anatomical follow-up at earlier stages and narrowed the window for diagnostic interventions or the option of legal abortion based on prenatal diagnosis.

Despite the diagnosis of posterior fossa malformation compatible with a variant of Dandy-Walker syndrome, the newborn presented a stable clinical course during the neonatal period. This case demonstrates that some variants of the Dandy-Walker spectrum can have a benign neonatal course, without signs of intracranial hypertension or evident neurological involvement at birth, reinforcing the importance of specialized postnatal follow-up. However, this outcome does not negate the need for early, multidisciplinary diagnosis beginning in the prenatal period.

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Conflict of interest

The authors declare that they have no conflicts of interest.

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Attachments

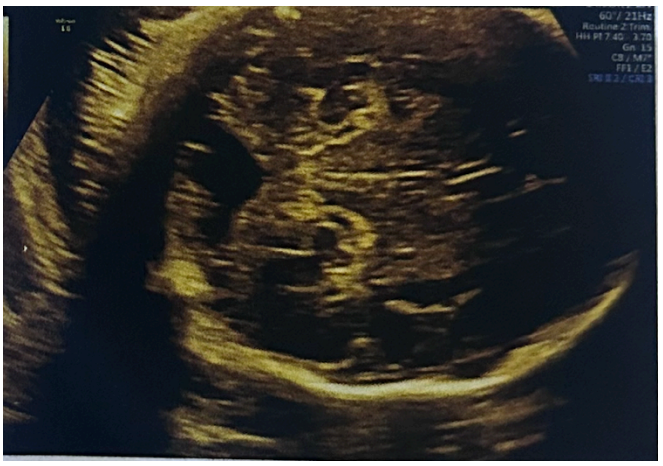

Figure 1. Transabdominal ultrasound, cross-section, shows absence of the cerebellar vermis at the level of the cerebellum, with dilation of the cisterna magna of 12.7 millimeters.

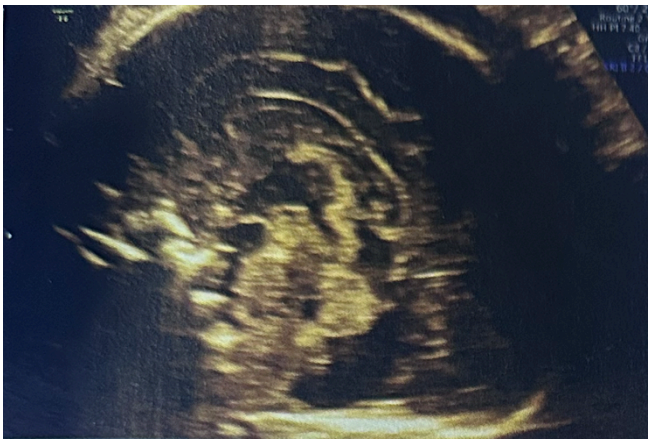

Figure 2. Transabdominal ultrasound: longitudinal section: complete absence of the cerebellar vermis, dilatation of the fourth ventricle and posterior fossa is evident.

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About the Authors

German Andres Guevara Lizarazo is a General practitioner from University of Pamplona and a Medical specialist in Epidemiology from the Universidad Autónoma de Bucaramanga. Currently, he works at the Gynecology and Obstetrics Service of The FOSCAL Clinic in Floridablanca.

Jonattan Palacios Torres is a general practitioner. He studied at the University of Sinú in Cartagena, Colombia. He works as a home hospitalization physician. He practices in the area of emergencies and surgery. Because of this, he has the training to work in hostile environments and high demand for emergencies.

Joshuart David Pascuas Varela is a physician who graduated from University of Magdalena in 2022 with more than 2 years of experience in surgical assistance, achieving multiple procedures alone and under guidance. He has been working in third and fourth level institutions in which the care of the critically ill patients is the priority. Among his core values we highlight assertive communication, responsibility, and broaden dedication to his patient's well-being. E-mail: Joshpamd@gmail.com.

Mayra Alejandra Vargas Quiroga is a general practitioner, graduated from the Universidad del Sinú, Cartagena, Colombia. She is a physician interested in specializing in Gynecology and Obstetrics. She is currently working in the Gynecology and Obstetrics Department of the FCV Cardiovascular Foundation in Floridablanca, Colombia. She has the experience to qualify for a specialization in this field. Email: mayravargas2701@gmail.com

Marbin Yulieth Alvarez Ramirez is a general practitioner graduated from the University of Santander. He works in the area of patient care in outpatient clinics, emergencies, and hospitalizations in a humane manner, empathizing with each of their pathologies and also caring for and protecting their mental health. He puts a great emphasis and interest in the area of gynecology, which is the area in which he wishes to specialize, ensuring the care and health of the mother-child binomial. E-mail: alvarez.yulieth@yahoo.com.