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Antenatal Diagnosis of Dandy-Walker Malformation Associated with Trisomy 13: A Case Report

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Abstract **Case Report**

Introduction: Dandy-Walker malformation (DWM), characterized by vermian hypoplasia, cystic dilatation of the fourth ventricle, and elevated tentorium cere- belli, is a rare cerebellar anomaly often associated with chromosomal disorders like trisomy 13. Antenatal ultrasound is critical for diagnosis. ***Case Presentation:*** A 20-week pregnant primigravida's fetus was diagnosed with trisomy 13 and DWM via trophoblast biopsy, presenting vermian agenesis, enlarged fourth ventricle, floating tentorium, and increased brainstem-tentorium angle on ultrasound. ***Results:*** Multidisciplinary evaluation confirmed the diagnosis, guiding counseling for a poor prognostic outcome. ***Conclusion:*** This case highlights the role of ultrasound in detecting DWM and trisomy 13, emphasizing the need for genetic testing and comprehensive counseling in complex fetal anomalies.

Keywords: Dandy-Walker malformation, trisomy 13, antenatal ultrasound, vermian agenesis, posterior fossa, genetic counseling.

1. INTRODUCTION

Dandy-Walker malformation (DWM) is a rare congenital anomaly of the posterior fossa, occurring in approximately 1/25,000–35,000 live births [1]. It is characterized by three key features: partial or complete agenesis of the cerebellar vermis, cystic dilatation of the fourth ventricle, and elevation of the tentorium cerebelli with an enlarged posterior fossa [2]. Trisomy 13, or Patau syndrome, is a severe chromosomal disorder with an incidence of 1/5000–20,000 live births, associated with multiple anomalies, including craniofacial, cardiac, renal, and central nervous system defects [3]. The association between DWM and trisomy 13 is well-documented, with 32.9 Antenatal ultrasound is the primary diagnostic tool for DWM, using axial and midsagittal views to assess the posterior fossa. Normal findings include a vermis more echogenic than cerebellar hemispheres, a triangular fourth ventricle, and a brainstem-tentorium (BT) angle of ¡30° [5]. In DWM, ultrasound reveals vermian hypoplasia, an enlarged fourth ventricle communicating with a posterior cyst, and an elevated tentorium (BT angle ¿45°) [2]. Trophoblast biopsy or amniocentesis confirms chromosomal anomalies, guiding prognostic counseling [6]. This case report describes the antenatal diagnosis of DWM in a fetus with trisomy 13, highlighting ultrasound findings, diagnostic criteria, and clinical management.

2. CASE PRESENTATION

A primigravida (G1P1) presented at 20 weeks and 4 days of gestation for evaluation following an abnormal ultrasound. Her pregnancy history and medical background were unremarkable, with no additional details provided on maternal health or obstetric history. Routine ultrasound at 13 weeks revealed suspected posterior fossa anomalies, prompting a trophoblast biopsy that confirmed a 46,XY karyotype with trisomy 13. Subsequent ultrasound at 20 weeks + 4 days, performed by a maternal-fetal medicine specialist, identified features consistent with Dandy-Walker malformation (DWM).

Axial ultrasound of the posterior fossa showed partial agenesis of the cerebellar vermis, an enlarged fourth ventricle with an abnormal crescent shape (concavity posterior), and an expanded great cistern. The cerebellar hemispheres appeared hypoplastic, and the Blake's pouch remnants were prominent, forming an oval-shaped cystic structure posterior to the vermis. Midsagittal ultrasound confirmed a floating tentorium

cerebelli, with an increased brainstem-tentorium (BT) angle of approximately 67° (normal ¡30°) and a brainstem-vermis (BV) angle of 45° (normal 0–10°). The fourth ventricle was dilated, communicating with a posterior fossa cyst, and the vermis was elevated, losing contact with the brainstem.

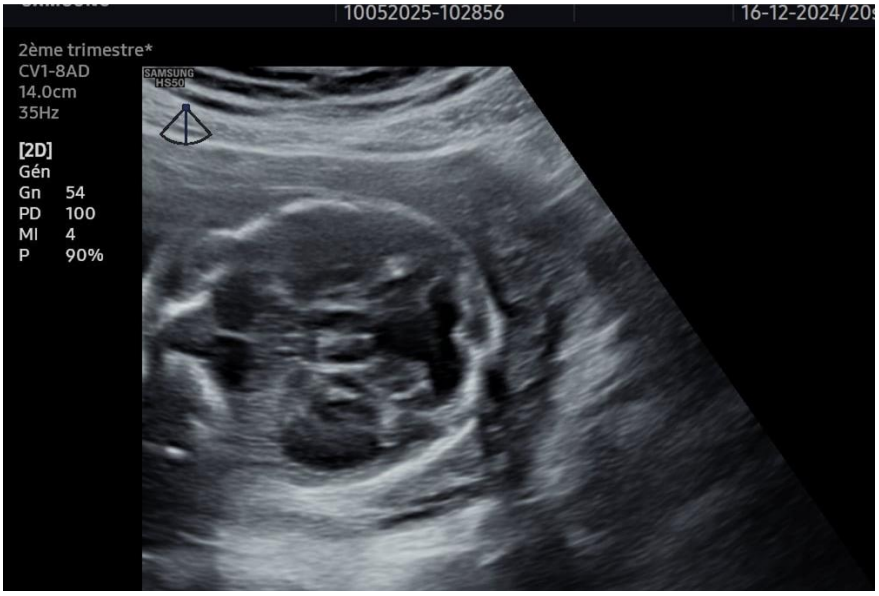

Figure 1: Axial ultrasound at 20 weeks showing partial vermian agenesis, enlarged fourth ventricle, and expanded great cistern

Additional ultrasound findings included normal supratentorial structures, with no evidence of hydrocephalus at this stage. The cavum septum pellucidum was visible, and the brainstem appeared intact. No other anomalies (e.g., cleft lip/palate, polydactyly) were reported in the presentation, though trisomy 13 of- ten presents multiple malformations. Fetal biometry and amniotic fluid volume were within normal limits. The patient was counseled by a multidisciplinary team, including maternal-fetal medicine, genetics, and neonatology, regarding the severe prognosis of trisomy 13 and DWM, which includes high neonatal mortality and profound neurodevel- opmental impairment [4]. The management decision (e.g., continuation or termination) was not specified in the presentation, but the context suggests preparation for a poor outcome.

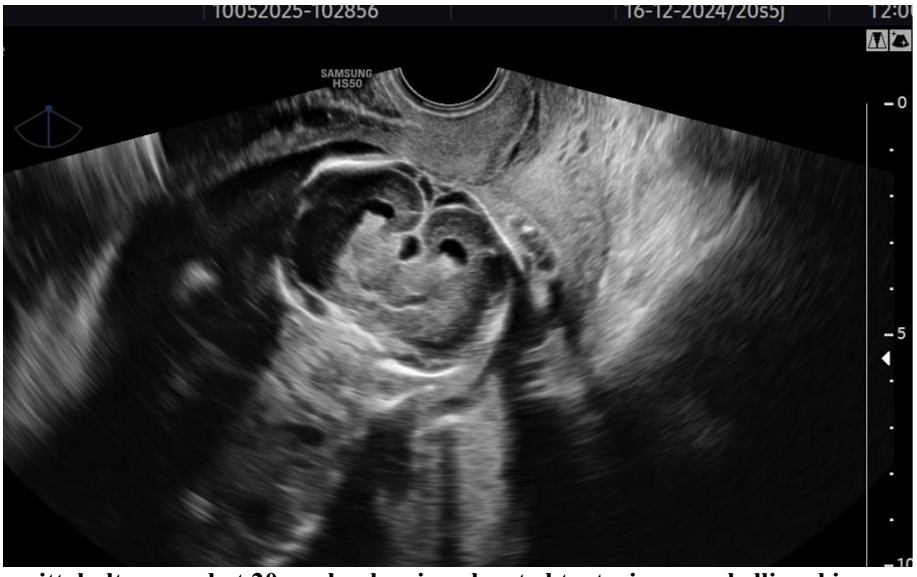

Figure 2: Midsagittal ultrasound at 20 weeks showing elevated tentorium cerebelli and increased brainstem-tentorium angle (67°)

3. DISCUSSION

Dandy-Walker malformation (DWM) is a spectrum of posterior fossa anomalies, classified by Guiboud in 2006 into three groups: increased retrocerebellar fluid (e.g., DWM, Blake's pouch cyst), reduced cerebellar biometry, and abnormal cerebellar anatomy [7]. This case falls within the first group,

characterized by vermian hypoplasia, cystic dilatation of the fourth ventricle, and an enlarged posterior fossa with elevated tentorium cerebelli [2]. Ultrasound is the cornerstone of diagnosis, with axial views showing an enlarged fourth ventricle and absent/vermian hypoplasia, and midsagittal views confirming an increased BT angle (¿45°) and vermian elevation [5]. In this case, the BT angle of 67° and BV angle of 45° were diagnostic of DWM, with the floating tentorium and cystic fourth ventricle clearly visualized.

The association of DWM with trisomy 13 is significant, with a study of 76 fetuses with DWM reporting chromosomal anomalies in 32.9

Differential diagnoses for DWM include Blake's pouch cyst, mega cisterna magna, and vermian hypoplasia without cystic dilatation [8]. Blake's pouch cyst, as noted in the presentation, features a normal vermis with an infravermian cyst communicating with the fourth ventricle, distinguishing it from DWM's vermian agenesis and elevated tentorium [7]. In this case, the partial vermian agenesis and increased BT angle ruled out Blake's pouch cyst. Fetal MRI, recommended for definitive diagnosis, was not reported but would have confirmed vermian elevation and cystic dilatation [9].

The prognosis of trisomy 13 with DWM is severe, with a median survival of 7–10 days and significant neurodevelopmental impairment in rare survivors [3]. A reported case of a 10-year-old girl with trisomy 13 and DWM highlighted exceptional survival but with severe neurological and gastrointestinal complications [4]. Management involves multidisciplinary counseling, weighing continuation versus termination based on parental values and prognostic data [10]. Ultrasound surveillance monitors progression (e.g., hydrocephalus), while genetic testing informs recurrence risk (typically ¡[1].

This case underscores the diagnostic utility of ultrasound, particularly midsagittal views for assessing the BT angle and vermian position, as emphasized by Guiboud's classification [7]. The increased BT angle (67°) and vermian agenesis were critical for distinguishing DWM from other posterior fossa anomalies. Future research should refine ultrasound criteria for early DWM detection and explore prenatal interventions, though the prognosis of trisomy 13 remains poor [9].

4. CONCLUSION

This case report describes the antenatal diagnosis of Dandy-Walker malformation in a fetus with trisomy 13 at 20 weeks' gestation, confirmed by trophoblast biopsy. Ultrasound findings of partial vermian agenesis, enlarged fourth ventricle, floating tentorium cerebelli, and increased brainstem-tentorium angle were diagnostic of DWM. The case highlights the association between DWM and trisomy 13, the critical role of axial and midsagittal ultrasound views, and the need for genetic testing to guide counseling. Clinicians should employ comprehensive ultrasound evaluation and multidisciplinary input to manage such complex fetal anomalies, ensuring informed decision-making for families facing a poor prognostic outcome.

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