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Prenatal Ultrasound Diagnosis of Dandy-Walker Malformation: About a Case and Review of the Literature

Ragmoun H, Mhelheli R, Hachicha S, Yahyaoui R and Hafsi M*

Department of Obstetrics and Gynecology, Menzel Temime Hospital, Tunisia

Abstract

Dandy-Walker syndrome is a heterogeneous group of cerebellar anomalies ranging from agenesis to hypoplasia or even early cerebellar atrophy. It can be isolated or associated with other malformations. The diagnosis can be made antenatally by morphological ultrasound in the second trimester or postnatally. The associated malformations and the degree of hydrocephalus are the two essential criteria that condition the subsequent prognosis.

We report a case of Dandy-Walker malformation that was diagnosed during a first routine prenatal ultrasound at 17 weeks of gestation. We will detail the contribution of ultrasound in the diagnostic process by comparing it with the data in the literature.

Keywords: Dandy walker malformation; Antenatal; Postnatal; Imaging

Introduction

Dandy-Walker syndrome is a group of heterogeneous cerebellar abnormalities starting from agenesis to hypoplasia or even early cerebellar atrophy.

It includes the Dandy-Walker malformation, the "Dandy Walker variant": Cystic dilatations of the V4 resulting from other types of vermian and/or cerebellar anomalies, which is a malformation that does not validate the 3 criteria of the real Dandy-Walker and the Magna Mega Cistern [1-3].

Before 18 weeks of pregnancy, the prenatal ultrasound diagnosis of the classic Dandy-Walker malformation is easier than the definitive diagnosis of "Dandy-Walker variant" and Mega-cisterna magna. A normal ultrasound appearance of the cerebellum in the second trimester does not rule out certain pathologies of the posterior fossa since its structures are not yet established. The sonographer must be careful before making a definitive diagnosis of "Dandy-Walker variant" or "mega-tank magna" [4].

We are reporting a case of Dandy-Walker malformation which was diagnosed during a first routine prenatal ultrasound at 17 weeks of pregnancy.

Case Presentation

This is a 29-year-old patient, gravida 3, para 2, married for 7 years, non-consanguineous marriage. In her obstetrical history, there is a vaginal delivery of a full-term newborn in good health and a C-section for fetal macrosomia with a good neonatal outcome.

She did a follow-up at 18 weeks of amenorrhea. Antenatal ultrasound performed at this term showed dolichocephaly with cystic malformation of the posterior fossa measuring 28 mm × 23 mm × 17 mm associated with agenesis of the cerebellum as well as significant hydrocephalus and agenesis of the corpus callosum found in sagittal section (Figure 1, 2). All of these conditions are consistent with Dandy-Walker malformation. In addition, there is an intrathoracic stomach hernia (Figures 1-3). An amniocentesis was performed showing a normal karyotype 46 XX. The case was presented in a multidisciplinary consultation meeting and a therapeutic interruption of the pregnancy was decided in agreement with the parents. Histopathological examination confirmed the ultrasound findings.

Discussion

Dandy-Walker malformation is a term that includes several anomalies of brain development. This anomaly is a congenital brain malformation usually involving the fourth ventricle and the cerebellum which was first described in 1914 by Dandy and Blackfan and was referred to as

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Correspondence:
Houssem Ragmoun, Department of Obstetrics and Gynecology, Menzel Temime Hospital, La Plage Street, Menzel Temime 8080, Tunisia, E-mail: houssem.ragm0@gmail.com

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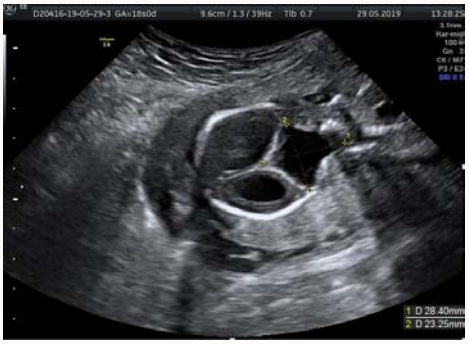

Figure 1: Trans-cerebellar coronal slice showing a cystic malformation of the posterior fossa with agenesis of the cerebellar vermis.

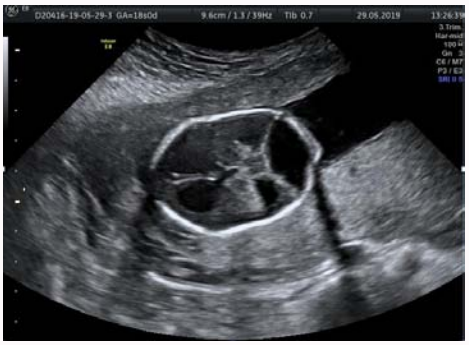

Figure 2: Cavo-thalamo-cerebellar section showing dolichocephaly with cystic malformation of the posterior fossa and agenesis of the cerebellar vermis.

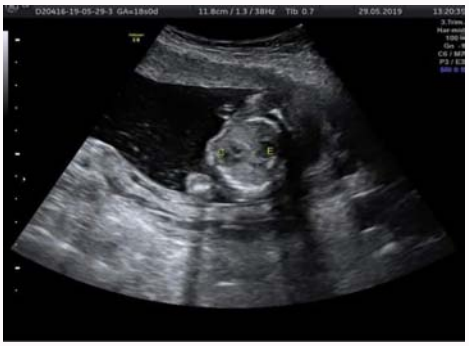

Figure 3: Axial thoracic section showing a right intrathoracic hernia of the stomach (E).

Dandy-Walker syndrome in 1954 by Benda, who also reported a familial origin [5]. The prevalence of Dandy-Walker malformation is approximately 1:30,000 live births with a slight female preponderance as is the case for our patient and is responsible for 4% to 12% of infantile hydrocephalus [6].

A bibliographic search on the "PubMed" and "ScienceDirect" platforms concerning the antenatal diagnosis of Dandy-Walker syndrome was carried out; three articles were found during the period of 2011-2022. The studies involved 8 cases in total.

The Dandy-Walker malformation is frequently associated with other intracranial abnormalities such as agenesis of the corpus callosum found in our case. Also, to be looked for: Holoprosencephaly, occipital encephalocele and ocular anomalies [7-9]. Extra-cranial anomalies are also to be sought and are in order of frequency: Cardiac anomalies (38%), facial dysmorphisms and cleft palate (26%), dysgraphia, poly- and syndactyly (28%), genital malformations-urinary (28%) and digestive (7%) [8].

Other less common abnormalities include limb abnormalities, abdominal wall abnormalities and diaphragmatic hernia which are extremely rare and found in our clinical case. Sexual ambiguities have also been described [10,11].

Postnatal studies indicate that the incidence of associated malformations is between 50% and 70% [12]. The most common etiology is genetic.

Environmental factors, infections, including viral (rubella, CMV), alcohol and diabetes also seem to play a role in the pathogenesis of Dandy-Walker malformation but the scientific evidence remains uncertain. In our patient, there does not seem to be any infectious, environmental or genetic factors, especially since the karyotype is normal. In the absence of an obvious cause, the risk of recurrence in subsequent pregnancies is 1% to 5% [12].

Prenatal ultrasound diagnosis of Dandy-Walker malformation is quite simple. However, it is important to differentiate with other posterior fossa anomalies such as mega cistern magna and retrocerebellar arachnoid cyst. For this, a set of ultrasound signs must be sought:

At the level of the Posterior Cerebral Fossa (PCF)

- In the cavo-thalamo-cerebellar section: A cystic dilation of the V4 leading to widening of the FCP, anechoic, triangular with an external base. the cerebellar hemispheres, hypoechogenic, are pushed back laterally and anteriorly, more or less hypotrophic, symmetrically or not. The vermis, echogenic, is hypotrophic or even absent (25%).

- In the sagittal section: The tentorium of the cerebellum is elevated; the vermis presents a more or less significant amputation of its lower part going as far as the complete absence of individualization. The lower part of the vermis can be extended towards the torcular by a linear image "in the tail of a comet" corresponding embryologically to the upper wall of the Blake pouch. The brainstem is normal.

On the supratentorial level

Anomalies are found in 70% of cases: Hydrocephalus (60%), agenesis of the corpus callosum (15%), migration anomalies, lipomas and interhemispheric cysts, encephalocele, holoprosencephaly [13].

As for the MRI, it finds its place especially in case of diagnostic doubt with the other cystic malformations of the posterior fossa or if there is strong presumption of a form close to the Dandy-Walker malformation with vermian hypoplasia formerly called "Dandy-Walker variant".

It not only helps to refine the diagnosis initially screened by ultrasound, but it can also represent a useful imaging technique for confirming and characterizing the various cerebellar malformations thanks to the median sagittal sections of the posterior fossa. At an early stage (19 to 24 weeks of gestation), it makes it possible to make the differential diagnosis between vermian malrotation and Dandy-Walker malformation by studying the biometry and morphology of all the structures involved [8].

Even in the second and third trimester, an inappropriate scan angle, can give the impression of excessive size of the cistern magna (great cistern) and even a vermian abnormality [14]. The retrocerebellar arachnoid cyst is a less frequent and more benign

anomaly than the Dandy-Walker malformation, because in this case, the overlying brain is normal. The cerebellar hemispheres are not separated by a cystic mass. Rather, they are moved en bloc [15].

As the congenital anomalies of the posterior fossa are numerous with an important anatomical nuance, a new ultrasound prenatal screening method was mentioned by an Italian-Polish team: It is the measurement of the angle of the Brainstem-Vermis (B-V) from 17 weeks of amenorrhea on the median sagittal slice. In this series, normal fetuses have an angle always less than 18°. For the Blake pouch cyst, this angle is less than 30°, unlike fetuses with a Dandy-Walker malformation which present an angle (B-V) always greater than 45°. An intermediate value is noted for vermian hypoplasia [16].

Early detection at 14 WA has been reported by measuring the angle (B-V) making it possible to perform a trophoblastic biopsy in search of chromosomal abnormalities [17].

Based on the available evidence, it is believed that prenatal ultrasound allows definitive diagnosis only of severe anatomical varieties of Dandy-Walker syndrome. These are characterized by both a large cystic mass in the posterior fossa and agenesis of the cerebellar vermis, which is called the classic Dandy-Walker malformation [18].

It is difficult to dispel doubt before birth about the presence of a mega-cistern magna or a small lower vermis anomaly. This can only be resolved by postnatal imaging studies (magnetic resonance imaging) or findings by fetal pathology study.

Conclusion

Dandy-Walker malformation is a rare and heterogeneous malformation involving the cerebellar vermis. The first ultrasound clue to the diagnosis is an enlargement of the cistern magna. Fetal MRI allows a better characterization of this malformation. This pathology is associated with CNS malformations and other genetic diseases, which is why a study of the karyotype and molecular analyzes are suggested. Prenatal diagnosis of DWM cannot predict the prognosis of the disease, on the basis of which the medical team and the parents must make a decision, especially since there is no consensus in the literature.

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