Case Study: Diagnosis of Dandy-Walker Malformation

These images of a fetal brain from a routine ultrasound examination at 22 weeks' gestation revealed a rare congenital brain abnormality.

This case involves a 34-year-old woman (gravida 3 para 2) who was 22 weeks pregnant. Before this pregnancy, her menstrual cycles were regular. She has reported no vaginal bleeding for 22 weeks, and she's had no clinically significant illnesses.

Clinical examination: The patient’s blood pressure was 124/82 mm Hg; her other vital signs also were normal. Chest examination was unremarkable, and the abdominal examination showed that the fundal height matched the gestational age of 22 weeks.

Imaging studies: This patient underwent a routine transabdominal obstetric ultrasound examination to exclude any congenital fetal abnormalities. She had not had any previous ultrasound scans related to this pregnancy. The resulting images are shown here.

Ultrasound Scan Findings
The most obvious finding on initial observation of these three images is a triangular defect towards the posterior aspect or occipital region of the fetal brain. The defect appears to involve the posterior aspect of the cerebellum. It does appear that there is adequate amount of amniotic fluid surrounding the fetal head. Also, the lateral ventricles seem to be normal in size. These findings imply several possibilities. The first possibility is a widened subarachnoid space, or prominent cisterna magna—the so-called mega-cisterna magna. For a diagnosis of posterior fossa lesions, however, the most important image is the sagittal section of the fetal brain.

Figure 4 and Figure 5, true mid-sagittal sections of the fetal brain, again show the triangular defect posterior to the cerebellum. The defect appears to be occupied by cerebrospinal fluid (CSF). But what has happened to the cerebellum and the posterior fossa?

The posterior fossa is definitely enlarged, and the cerebellum appears to be compressed. Also, the cerebellum is markedly hypoplastic, with almost no display of the vermis, a vital part of the cerebellum that connects the two lateral hemispheres. This is an important finding and changes the course of the diagnosis. Now, the possible differential diagnoses are arachnoid cyst, Blake pouch cyst, Arnold-Chiari malformation, and Dandy-Walker malformation.

An arachnoid cyst can be ruled out because what may be a cystic structure in the posterior fossa is in actuality the fourth ventricle. Also, the severe hypoplasia of the cerebellum and the vermis also help rule out a diagnosis of an arachnoid cyst.

Next, Blake pouch cyst can be ruled out because the cerebellar vermis would be compressed and elevated, not severely hypoplastic as it is in this case.

An Arnold-Chiari malformation is not likely in this case, as this would cause an effacement of the cisterna magna and the fourth ventricle, unlike the findings seen here.

This leaves Dandy-Walker malformation. To recap the findings, there is an enlarged posterior fossa, cystic dilatation of the fourth ventricle, no significant dilatation of the lateral ventricles, near absence or hypoplasia of the cerebellum and the vermis, and elevation of the tentorium cerebelli. Together, these findings match the signs of Dandy-Walker malformation.

Final diagnosis: Dandy-Walker malformation

Discussion
Dandy-Walker malformation was first coined to describe the congenital anomaly of a cystic dilatation of the fourth ventricle along with partial or complete agenesis of the vermis of the cerebellum. Subsequently, medical literature identified four classic signs of this malformation: complete or partial agenesis of the vermis of the cerebellum; cystic enlargement of the fourth ventricle; enlargement of the posterior fossa; and cranial displacement of the tentorium cerebelli with anti-clockwise rotation of the vermis. The etiology of this malformation is based on the finding that there is a defect in the development of the roof of the fetal rhombencephalon along with a partial or complete lack of patency of the foramen of Magendie.
Prognosis
The effect of Dandy-Walker malformation (also called Dandy-Walker Syndrome) on intellectual development is variable. Some children have normal cognition, and others never achieve normal intellectual development even with early drainage of excess CSF buildup. Longevity depends on the severity of the malformation and whether other associated malformations are present. For the fetus in this case, the prognosis is very poor because of the severe hypoplasia of the vermis and the extreme enlargement of the fourth ventricle. In such cases, the chances of severe enlargement of the lateral ventricles are very high during the later stage of pregnancy. Hence, the mortality rate in such fetuses both during pregnancy and in the immediate postnatal period is high. Fetuses that survive to the postnatal period are likely to have severe cognitive impairments and are at risk for damage to the cerebrum during infancy.

Disclosures:
The images in this case are courtesy of Firoz Bhuvar, MD.

References:

Paladini D, Volpe P. Central and peripheral nervous system anomalies. In: Ultrasound of Congenital Fetal Anomalies: Differential Diagnosis and Prognostic Indicators (Series in Maternal Fetal Medicine).

Taylor and Francis Group LLC; Boca Raton, Fla; 2007:11-62.


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