Prenatal Diagnosis of Herniated Dandy-Walker Cysts

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Objective. The purpose of this series is to describe the prenatal diagnosis and pregnancy outcome of fetuses affected with Dandy-Walker malformation in which a posterior cyst herniated through a bony defect of the occipital skull, foramen magnum, or both. Methods. Two- and 3-dimensional sonography were used to examine 2 fetuses with poorly delineated cerebellar structures and a large posterior cystic neck mass. Fetal magnetic resonance imaging (MRI) was added to this evaluation as a complementary diagnostic modality. Results. Three-dimensional sonography helped characterize the precise site of cyst herniation through the occipital skull or foramen magnum. Fetal MRI confirmed the sonographic findings. Neonatal MRI studies identified heterotopic gray matter as evidence of a neuronal migration disorder in both fetuses. The second fetus also had agenesis of the corpus callosum. Conclusions. The antenatal detection of a large posterior cystic neck mass and a poorly defined or nonvisualized cerebellar vermis suggest Dandy-Walker malformation with a herniated cyst. Three-dimensional sonography and fetal MRI are important adjunctive methods that can be used to evaluate the herniation site and a possible neuronal migrational disorder. Key words: Dandy-Walker malformation; fetal magnetic resonance imaging; herniated cyst; heterotopia; 3-dimensional sonography.
Case Descriptions

Case 1
The patient was a 33-year-old primigravida who was referred for the evaluation of fetal growth at 18.6 weeks’ menstrual age (MA). Her maternal serum α-fetoprotein level was normal. A cystic posterior neck mass, measuring 2.5 × 2.7 cm, was present with intact overlying skin (Figure 1). No septations or solid components were identified. The cerebellum and cisterna magna were poorly visualized. The detailed scan and echocardiographic findings were otherwise normal. Fetal biometric values were normal. These findings suggested an occipital meningocele with possible involvement of the cervical spine. A normal fetal karyotype (46,XX) was obtained by amniocentesis. By 23.9 weeks, the cystic neck mass was associated with mild enlargement of the lateral and third brain ventricles. Again, the cerebellar hemispheres and cisterna magna were poorly visualized. Three-dimensional sonography provided multiplanar views and clear identification of a bony skull defect by volume rendering (Figures 2 and 3). An occipital meningocele with an Arnold-Chiari malformation was suspected, although the possibility of a DWM was also considered.

At 37.4 weeks, the patient underwent a spontaneous vaginal delivery. A 3094-g female neonate was delivered without complications (Figure 4). Neonatal MRI identified a DWM with marked cerebellar hypoplasia and partial absence of the cerebellar vermis. A suboccipital fluid-filled cyst (6.3 × 4.5 × 8.4 cm) directly communicated with the enlarged cisterna magna (5.0 × 4.8 × 3.3 cm).

The cyst extended through a midline defect of the occipital skull that was contiguous with a widened foramen magnum. Subependymal nodules in the body and trigones of the lateral ventricles indicated the presence of heterotopic gray matter (Figure 5). Focal hemorrhage was present in both posterior parietal lobes. The final diagnosis was a herniated Dandy-Walker cyst with a migrational brain disorder and possible venous infarction.

At 11 months after delivery, the infant could not sit independently and had no eye tracking. There was generalized hypotonia with upper limb spasticity. This was complicated by swallowing difficulties that made tube feedings necessary. Periods of apnea were also noted, and death occurred at approximately 1 year of age.

Case 2
A 25-year-old primigravida was referred for fetal growth evaluation at 18.3 weeks’ MA. A cystic mass (3.4 × 2.5 cm) protruded from the occipital skull and appeared to be connected to the posterior fossa (Figure 6, A and B). No spinal abnormalities were visualized. Bilateral frontal bone scalloping (“lemon sign”) was present. Agenesis of the corpus callosum was suspected on the basis of a nonvisualized septum pellucidum, narrowed anterior horns, and colpocephaly (Figure 6C). The cerebellum initially had a normal appearance. A herniated DWM and agenesis of the corpus callosum were suspected. The possibility of an occipital meningocele could not be excluded. Amniocentesis revealed a normal fetal karyotype. By 25.9 weeks, the cerebellar hemispheres...
were more obviously splayed, and the fourth ventricle was clearly connected to an enlarging cystic mass (Figure 6D). The site of cyst herniation was clearly delineated by 3-dimensional sonography (Figure 7). Fetal MRI confirmed the extrusion of the large cyst through the foramen magnum (half-Fourier acquisition single-shot turbo spin echo; Sonata 1.5 T; Siemens Medical Solutions, Mountain View, CA).

The patient gave birth to a 4575-g female neonate by primary classic cesarean delivery at 37.0 weeks. Neonatal MRI revealed a large herniated cyst, from the low occipital region and foramen magnum, with agenesis of the corpus callosum (Figure 8). Partial absence of the cerebellar vermis indicated an incomplete DWM. The postnatal MRI scan showed subependymal nodules that were retrospectively correlated to the fetal MRI scan as well (Figures 9 and 10). Retrospective review of 3-dimensional sonographic volume data sets also revealed heterotopic gray matter at 18.7 weeks’ MA (Figure 11). The cystic mass was surgically excised, and an arachnoid defect was repaired on the third postpartum day. Postoperatively, hydrocephaly developed, and the neonate required placement of a ventriculoperitoneal shunt before discharge on day 15. No focal neurologic deficits or seizures had been noted by 6 months of age.

Figure 2. Case 1. Three-dimensional multiplanar imaging (Echoscan; TomTec Imaging Systems, Munich, Germany) showed spatial relationships of the neck mass to other anatomic structures in orthogonal scanning planes.

Figure 3. Case 1, 3-dimensional volume reconstructions of the cystic neck mass (Echoscan; TomTec Imaging Systems). A posterior angled view of the neck mass (left image) was digitally exposed to show cyst herniation through the foramen magnum (right image, asterisk).
Discussion

Dandy-Walker malformation can develop as a separate entity or in association with genetic syndromes, chromosomal aberrations, and congenital infections. During embryogenesis, the area membranacea appears as a rudimentary structure that normally separates the developing cerebellar vermis from the primitive choroid plexus. This membranous area is normally incorporated into the choroid, unless it expands into a dilated cystic mass of the fourth ventricle as part of the DWM.

The prenatal sonographic diagnosis of congenital DWM is usually made from an enlarged posterior fossa cyst, splayed cerebellar hemispheres, and an incompletely formed cerebellar vermis. These abnormalities are typically recognized during mid gestation, although the diagnosis has been made as early as 13 weeks’ MA on transvaginal sonography. Occasionally, DWM can be difficult to distinguish between other posterior fossa abnormalities such as the Dandy-Walker variant, arachnoid cyst, and megacisterna. Partial development of the cerebellar vermis can also occur as part of a Dandy-Walker variant. The vermis, however, may not be completely developed until after 18 weeks’ MA.

Our findings show an unusual early variation of DWM in which a posterior cyst actually herniates through the foramen magnum, a contiguous occipital skull defect, or both. Three-dimensional sonography and fetal MRI were used to precisely show the herniation site and to identify heterotopic gray matter as prenatal evidence of a migrational brain disorder. We are aware of only 1 other case report that presents the sonographic appearance of a herniated “meningocele” with DWM in a fetus at 35 weeks’ MA. This fetus had a diagnosis of hydrocephaly and subsequently had delayed psychomotor development by 7 months of age. Goldstein et al also described 2 fetuses with DWM and cephalocele, although no images were presented.

Occipital cephaloceles with DWM have been primarily described in the pediatric literature. Bindal and colleagues reported 8 (16%) of 50 infants with occipital meningoceles and DWM. All infants had a communication between the occipital meningocele and posterior fossa. Although 2 of their cases were diagnosed prenatally, no fetal sonographic images were presented. Outcome was related to the presence of other findings such as schizencephaly, agenesis of the corpus callosum, and the presence of heterotopic gray matter. Only 3 infants were reported to have normal intelligence. Both of our fetuses had postnatal confirmation of additional brain abnormalities such as agenesis of the corpus callosum and heterotopic gray matter.

Identification of fetuses at risk for migrational disorders of cortical brain development can greatly affect antepartum counseling. The prenatal detection of nodular heterotopia by 2-dimensional sonography or fetal MRI has been
described as early as 23 weeks’ MA. Recent reports also suggest that either abnormal sulcal development\textsuperscript{18} or, possibly, the early presence of abnormally formed sulci and gyri\textsuperscript{19} are important diagnostic markers for migrational disorders. In our study, a retrospective examination of videotaped sonographic scans did not identify evidence for a migrational disorder in either case. However, a systematic review of 3-dimensional multiplanar images from case 2 provided the means for showing ventricular wall nodularity that was consistent with MRI studies. Prenatal identification of a cortical migrational disorder can be maximized by considering gestational age, using an endovaginal probe for cephalic presentation, manipulating the fetal head for an acoustic window through a fontanelle or suture, and remembering to apply the highest probe frequency for the best image resolution.

In summary, the antenatal detection of a large posterior cystic neck mass with a poorly defined or nonvisualized cerebellar vermis suggests DWM with a herniated cyst. These findings warrant a careful search for additional anomalies. Three-dimensional sonography and prenatal MRI are important adjunctive methods that can be used to evaluate the herniation site and the possibility of neuronal migrational disorders. A detailed evaluation for associated anomalies will assist clinicians who seek to counsel parents about pregnancy options, antenatal care, and prognosis for affected infants.

Figure 6. Case 2, representative sonographic views of a fetal head on conventional sonography. A, Posterior nuchal cystic mass (20.7 weeks’ MA). B, A bony defect of the occipital skull was suspected (asterisk, 19.7 weeks’ MA). C, Narrowed anterior ventricular horns with colpocephaly that suggested agenesis of the corpus callosum were revealed (30.7 weeks’ MA). D, An enlarged posterior fossa (asterisk) was revealed that was contiguous with a widened fluid-filled space between the cerebellar hemispheres (30.7 weeks’ MA). The findings were consistent with incomplete DWM due to abnormal development of the cerebellar vermis; ant indicates anterior; post, posterior; and sp, spine.
Figure 7. Case 2, 3-dimensional multiplanar views of the fetal head at 18.3 weeks’ MA (Voluson 730 Expert; GE Healthcare, Milwaukee, WI). A horizontal green line (top panels) represents the cutting plane through which the rendered volume reconstruction was generated to show the herniation site (bottom right panel).

Figure 8. Case 2. The neonate had a large posterior cystic mass (left) that extruded through a low occipital skull defect and widened foramen magnum as shown on the sagittal spin echo T1-weighted image (right).

Figure 9. Case 2. An axial view of the neonatal MRI (turbo spin echo T2-weighted image) showed nodules (arrows) from the presence of heterotopic gray matter. Tapered anterior ventricular horns and colpocephaly are important diagnostic markers for agenesis of the corpus callosum.
Figure 10. Case 2. Heterotopic gray matter nodules were identified on both prenatal (25.9 weeks’ MA) and postnatal MRI scans. The fetal MRI (left image, sagittal view, half-Fourier acquisition single-shot turbo spin echo sequence) showed low-signal nodules (asterisks) along the contours of the posterior lateral ventricle. Corresponding high-signal nodules (asterisks) were found on the neonatal MRI study from a similar but not identical sagittal view (T1-weighted image, right).

Figure 11. Case 2, multiplanar view of the fetal head and posterior cystic neck mass at 18.7 weeks’ MA. A sagittal view showed nodular projections (arrows) along the occipital ventricular wall, suggesting the presence of heterotopic gray matter.
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References


