First Trimester
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Articles in this Collection


Placental Mesenchymal Dysplasia (PMD) in Association With Beckwith-Wiedemann Syndrome Identified by First Trimester Sonography

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Abstract
Placental mesenchymal dysplasia (PMD) has commonly been identified on second trimester ultrasound in association with Beckwith-Wiedemann syndrome (BWS). In this report, a case of PMD later confirmed as Beckwith-Wiedemann is presented, which was identified by sonography in the first trimester. When faced with a first trimester finding of an enlarged cystic placenta, it is suggested that BWS be considered as a possible diagnosis and accordingly, genetic testing with methylation studies offered to the parents.

Keywords
Beckwith-Wiedemann, placental mesenchymal dysplasia, first trimester, sonography

Introduction
Placental mesenchymal dysplasia (PMD), known also as mesenchymal stem villous hyperplasia, is a rare placental vascular abnormality of unknown etiology with a reported incidence of 2 in 1000 cases.¹² Due to its relatively new recognition, there is paucity of data with respect to outcome of affected pregnancies. Nevertheless, recent reports, including a systematic review by Nayeri and colleagues,³ have implicated an association with Beckwith-Wiedemann syndrome (BWS) and adverse perinatal outcomes including intrauterine growth restriction (IUGR) and intrauterine fetal demise (IUFD). The sonographic presentation may demonstrate an enlarged, hydropic, and multicystic placenta. The cysts are typically well defined and may be separated by placental tissue, as opposed to a placental mass primarily comprised of cysts, as may be seen with gestational trophoblastic disease. The fetal anatomy will likely be unremarkable except in cases of associated Beckwith-Wiedemann syndrome.² Long-term outcomes are favorable so long as the fetus survives any complications stemming from IUGR and related pathology.

Beckwith-Wiedemann syndrome, which can be due to uniparental disomy of 11p15, is associated with the sonographic findings of macroglossia, macrosomia, organomegaly, abdominal wall defects, and placentomegaly.⁴ Fetuses diagnosed with BWS may have favorable outcomes if they survive the neonatal period, although they may have difficulty with feeding or breathing due to macroglossia. Mental status may be normal or slightly reduced. Other possible long-term complications include development of seizures, hypoglycemia, and embryonal tumors such as neuroblastoma and hepatoblastoma.⁴ Although recently linked with PMD, the reports have predominantly included second/third trimester or postpartum diagnoses. In the following we present a first trimester case of PMD associated with BWS.

Case Presentation
A 32-year-old gravida 2, para 0 Asian woman was referred to the clinic for a routine first trimester risk assessment and nuchal translucency measurement at 13 weeks and 3 days gestation, based on her last menstrual period. A sonographic examination was performed using a Voluson E8 machine (GE Healthcare, Fairfield, Connecticut, USA). Sonography showed that the fetus demonstrated a normal early anatomic survey and nuchal translucency...
measurement. However, the placenta appeared enlarged, hydropic, and contained numerous small cystic appearing areas (Figure 1), consistent with PMD. Prenatal diagnostic testing of chorionic villous sampling (CVS) was offered to the patient, which she accepted. The genetic evaluation demonstrated normal 46,XY karyotype and microarray; however, the methylation assay for Beckwith-Wiedemann syndrome proved positive. On follow-up examination (at 16 weeks) to discuss the results of the genetic testing, an isolated small omphalocele was also noted (Figure 2). After genetic counseling and consideration of their options, the couple elected to terminate the pregnancy. Post-delivery gross examination confirmed the presence of an omphalocele along with macroglossia.

**Discussion**

Prenatal sonographic findings of placental mesenchymal dysplasia and an omphalocele in association with Beckwith-Wiedemann syndrome are well documented in the current literature. However, these findings are predominantly described in the second or third trimesters of pregnancy. Although placental mesenchymal dysplasia has been reported in the first trimester, a literature search showed no reports of first trimester cases in association with BWS, and this is the first reported case.

PMD is underdiagnosed and underreported by many providers, including sonologists, as it is a relatively new entity first described in 1991 by Moscoso et al., and many remain unfamiliar with its presentation. PMD is often mistaken for gestational trophoblastic disease due to the similarities between the two entities with respect to the placental findings. The well-described first trimester sonographic findings associated with PMD include enlarged placenta, numerous cysts with placental tissue visible between them, and dilated chorionic vessels (Figures 3 and 4). The fetus may be normal but should undergo targeted sonography for signs of IUGR and BWS. Sonographically, the appearance of a cystic placenta may be similar to that of gestational trophoblastic disease. However, in gestational trophoblastic disease the placentae mass is often described as hypervascular by color Doppler sonography. In addition, the cystic components may not be well circumscribed and can be so numerous they essentially replace all placental tissue. With molar pregnancy there may not be a fetus at all (complete mole), or if a fetus is identified there may be signs of triploidy (IUGR, hydrocephalus, syndactyly, oligohydramnios) as opposed to the aforementioned signs of a fetus with BWS. It can be difficult to discern a twin pregnancy with a complete mole and a coexistent normal twin. In this case, it is important to identify the placenta of the normal twin to help distinguish from PMD. Although BWS is associated with presence of an omphalocele (Figure 5), this often may not be diagnosed in the first trimester as there may be physiologic fetal gut herniation. Furthermore, macroglossia and organomegaly are also features, which
Figure 2. Color Doppler image taken at 16 weeks gestation showing the omphalocele.

Figure 3. An example of placental mesenchymal dysplasia at 20 weeks gestation showing an enlarged placenta with numerous small cysts and dilated chorionic vessels.
present later in gestation, all of which underscore the importance of using PMD as a clue for BWS investigation. When faced with the aforementioned sonographic findings, the sonologist should evaluate the pregnancy for the following differential diagnoses: molar pregnancy (often accompanied by markedly elevated
human chorionic gonadotropin levels), chorangioma (often well circumscribed and vascular), subchorionic hematoma (often localized to base of placenta and not the parenchyma), and hydropic placentation (often related to an underlying infection or fetal isoimmunization). Karyotype should be offered to rule out molar gestation and methylation studies done to assess for BWS. An infectious disease panel should also be carried out.

Conclusion

The first known report of a first trimester case of placental mesenchymal dysplasia in association with Beckwith-Wiedemann syndrome is presented. When faced with a first trimester finding of an enlarged cystic placenta, it is suggested that BWS be considered as a possible diagnosis.

Declaration of Conflicting Interests

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References

Case Study

First Trimester Diagnosis of Omphalocele: Differentiating Between Omphalocele and Normal Physiologic Gut Herniation

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Abstract
Normal physiologic herniation of the fetal bowel is often seen on first trimester sonogram. An omphalocele is an abdominal wall defect containing bowel and/or liver and occurs as a result of herniation into the umbilical cord and is often associated with chromosomal anomalies. This case report presents an omphalocele seen in the first trimester and discusses how to differentiate this finding from normal physiologic bowel herniation.

Keywords
first trimester, omphalocele, midgut herniation, sonography

Discussion
Normal embryologic maturation of the abdominal wall and bowel occurs during the fourth week of gestational age. This process involves three folds: foregut, midgut, and hindgut. The foregut makes up the esophagus, stomach, duodenum, liver, pancreas, biliary tract, pharynx, and lower respiratory tract. The midgut structures include the small intestine, cecum, appendix, ascending colon, and the proximal portion of the transverse colon. The hindgut includes the distal portions of the alimentary tract and parts of the genitourinary tract. In the early stages the small bowel tends to grow quickly and outpace the capacity of the abdominal cavity, thus protruding into the base of the umbilicus (Figure 3). Abnormalities are frequently seen during this midgut fold formation.1-3 In this stage, the intestines will herniate into the umbilicus, twist 90°, and conclude with a 180° counterclockwise twist before descending back into the abdominal cavity. In some instances, there is incomplete rotation of the bowel, which leads to an omphalocele.

An omphalocele is an encapsulated abdominal wall defect containing bowel and/or liver and occurs as a
result of incomplete rotation of bowel returning into the abdominal cavity (Figure 4). An omphalocele is not typically an isolated finding.\(^7\)\(^9\) Concomitant with an omphalocele are a plethora of potential abnormalities including congenital heart defects (47%), genitourinary defects (40%), and neural tube defects (39%).\(^6\)\(^1\) Intrauterine growth restriction has also been found to be associated with an omphalocele.\(^1\)\(^2\) The main chromosomal defects that would be found with an omphalocele are trisomy 13 (Patau syndrome) and trisomy 18 (Edward syndrome).\(^4\) Due to the coexistence of multiple anomalies, a fetus found to have an omphalocele has a high mortality rate.\(^1\)\(^2\)

A number of significant features differentiate physiologic midgut herniation from an omphalocele (Table 1): (a) Midgut herniation descends back into the abdominal cavity by 12 weeks gestational age. (b) Midgut herniation will have a heterogeneous appearance within the encapsulated umbilicus, while an omphalocele will tend to have a more homogeneous appearance. (c) Midgut herniation will present as a small spherical shape, while an omphalocele will present with a large circular shape anterior to the abdominal wall. (A physiologic midgut herniation measures between 4 and 7 mm [Figure 5], and an omphalocele measures greater than 7 mm.\(^1\)\(^0\)) (d) The “tip of the iceberg” sign for an omphalocele is the definite appearance of the liver along with small bowel within the herniation.

Sonography can play a significant role in discerning midgut herniation from an omphalocele. Careful first trimester scanning of the anterior abdominal wall is crucial. It is important to determine the gestational age of the fetus being scanned and determine whether a physiologic midgut herniation should be present. The sonographer must examine the echogenicity of the herniation at the base of the umbilicus and make a determination as to heterogeneity or homogeneity. If the sonographer concludes that the herniation is too large to be considered a physiologic midgut herniation, measurements must be taken at the base of the umbilical herniation. Since an omphalocele is not usually an isolated finding, a survey...
appropriate to gestational age should be used to further locate other potential abnormalities, with a follow-up anatomical survey when more anatomy can be identified. An omphalocele is often found with an increased nuchal translucency (>3 mm) on the back of the fetal neck (Figure 1). Another common finding is fetal intrauterine growth restriction (fetal weight <10th percentile).12

Upon diagnosis of an omphalocele via sonography, it is usually recommended that the mother receive a chromosomal screening analysis. The chromosomal analysis can be accomplished by either a blood test, such as cell-free DNA, or an amniocentesis.1,5 Amniocentesis will provide a definitive diagnosis of a wide variety of congenital conditions, including trisomy 13 and trisomy 18.

The prognosis of a fetus with an omphalocele is not always lethal. More often than not it ends in a fetal demise because of the associated chromosomal abnormalities.4,13 However, there are occurrences where a fetus progresses to full-term birth and is delivered. Upon delivery, surgery is performed on the fetus’s abdominal wall to correct the omphalocele and place the intestines back into the abdominal cavity.3,14 Studies have been performed to determine whether postbirth repair is an option. Montero et al15 suggested using the omphalocele diameter in comparison to the abdominal circumference, femur length, and head circumference to evaluate whether the omphalocele is compatible for repair. With a ratio of 0.21 or greater, it is considered a “giant” omphalocele, which results in a poor outcome.

Conclusion

Midgut herniation is a normal physiologic occurrence during fetal development. However, complications can arise during this phase of fetal development, including an omphalocele. An omphalocele is an abdominal wall defect containing bowel and/or liver as a result of herniation into the umbilical cord. This abnormality is often associated with chromosomal anomalies, most commonly trisomy 13 and trisomy 18. With attentive and thorough scanning, an omphalocele can be detected and differentiated from normal physiologic herniation, which has a significant effect on the outcome in clinical decision making.

Declaration of Conflicting Interests

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References


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1. What placental characteristic is not associated with placental mesenchymal dysplasia (PMD)?
   A. Enlarged placenta
   B. Multicystic placenta
   C. Atrophic placenta
   D. Hydropic placenta

2. As indicated in the article, what is the reported incidence of PMD?
   A. 2 in 100 cases
   B. 1 in 200 cases
   C. 1 in 2000 cases
   D. 2 in 1000 cases

3. What statement concerning fetuses diagnosed with Beckwith-Wiedemann Syndrome is not true?
   A. Mental status may be slightly reduced
   B. They may develop seizures
   C. They always have a poor prognosis
   D. They may develop embryonal tumors

4. PMD has been commonly identified on second trimester ultrasound in association with:
   A. Trophoblastic disease
   B. Ectopic pregnancy
   C. Trisomy 13
   D. BWS Syndrome

5. What statement concerning omphalocele is not true?
   A. Omphalocele is typically an isolated finding
   B. An omphalocele is an encapsulated abdominal wall defect
   C. An omphalocele results from the incomplete rotation of bowel returning into the abdominal wall cavity
   D. The omphalocele typically contains bowel and/or liver

6. During what week of gestation does normal embryologic maturation of the abdominal wall and bowel occur?
   A. The 2nd week
   B. The 4th week
   C. The 6th week
   D. The 8th week

7. Which characteristic of an omphalocele is not true?
   A. An omphalocele presents with a large circular shape anterior to the abdominal wall
   B. The appearance within the encapsulated umbilicus will typically have a heterogeneous appearance
   C. The omphalocele may have a “tip of the iceberg” sign
   D. Possible ascites may occur within the membrane