Isolated echogenic bowel diagnosed on second-trimester ultrasound

Q. A 33-year-old G2 P1 patient at 18 weeks’ gestation presents to the ultrasound unit for a routine anatomic survey. Her medical and family histories are unremarkable and her pregnancy has been uncomplicated. A finding of isolated fetal echogenic bowel is noted. Fetal growth is appropriate for gestational age and no other anomalies are detected. How is the diagnosis of echogenic bowel made and how common is this finding on second-trimester ultrasound?

A. Echogenic bowel is a nonspecific finding observed during 0.2% to 1.8% of routine second-trimester ultrasound exams. Diagnosis is made when the fetal bowel displays an echogenicity or brightness equal to or greater than that of surrounding fetal bone, typically the iliac wing. If echogenic bowel is suspected, the ultrasound gain should be turned down to the lowest setting at which bone still appears white (Figure 1). If the fetal bowel continues to have an echogenic appearance, the diagnosis can be made.1

Transducer frequency also can affect the diagnosis because higher-frequency transducers tend to exaggerate the finding secondary to enhanced visualization of the small bowel submucosa.2 Therefore, the diagnosis of fetal echogenic bowel should be routinely confirmed at the lowest gain and with a lower-frequency transducer, typically 5 MHz or less. Despite these measures, the diagnosis of echogenic bowel continues to have a subjective component.

Although a grading system of degree of echogenicity has been proposed, bowel not as bright as bone has not been associated with an increased risk of adverse outcomes.3

What is the differential diagnosis of fetal echogenic bowel observed on second-trimester ultrasound?

Although echogenic bowel can be a transient, idiopathic finding in approximately 0.5% of fetuses, it also can be associated with a wide range of pathologic conditions such as primary gastrointestinal pathology, aneuploidy, cystic fibrosis, and congenital viral infection (See “Differential diagnosis for most common etiologies for echogenic bowel”). The estimated incidence of these conditions is highly variable, likely due, in part, to the relatively small sample sizes studied and the subjective component of the diagnosis of echogenic bowel.

In fetuses with a nonpathological cause for echogenic bowel, the primary mechanism is thought to be accumulation with meconium.1 Prior studies also have demonstrated the development of echogenic bowel following invasive procedures such as amniocentesis and intrauterine fetal transfusions secondary to fetal swallowing of blood from the amniotic cavity. It has been demonstrated that this finding may even persist for 2 to 4 weeks following the procedure.4,5 Primary gastrointestinal pathology such as bowel obstruction, atresia, and perforation also may cause an echogenic appearance of the fetal bowel. In cases of obstruction and atresia, decreased meconium fluid content is the proposed cause for the increase in echogenicity; however, the presence of meconium outside the intestinal lumen likely is responsible for the echogenic appearance in cases of bowel perforation.1,3 Echogenic bowel also has been reported in cases of Hirschsprung disease.6

The estimated incidence of aneuploidy in fetuses with isolated echogenic bowel ranges from 3.3% to 16%, with trisomy 21 being the most commonly diagnosed aneuploidy in this population.5,11 Other karyotypic abnormalities, such as trisomy 18, trisomy 13, Turner syndrome, and chromosomal mosaicism, also have been reported.17 Hypoperistalsis due to mechanical or functional bowel obstruction with subsequent dehydration of meconium is
Prenate®
Rx prenatal vitamin & DHA

DESCRIPTION: Prenate® ESSENTIAL™ is a prescription prenatal/vitamin/mineral/essential fatty acid softgel. Each softgel is blue-green in color, opaque, and impregnated with "Prenate®" on one side.

Supplement Facts
Serving Size 1 Softgel

<table>
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<th>Amount Per Serving</th>
<th>% DV For Adults</th>
<th>% DV For Pregnant and Lactating Women</th>
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Other Ingredients: Fish oil, gelatin, hydrogenated vegetable oil, glycerin, sorbitol, beeswax, soy lecithin, titanium dioxide, vanillin, FD&C blue No. 1, propylene glycol, hypromellose.

INDICATIONS: Prenate® ESSENTIAL™ is a multi-vitamin/mineral/essential fatty acid nutritional supplement indicated for use in improving the nutritional status of women throughout pregnancy and in the postpartum period for both lactating and non-lactating mothers. Prenate® ESSENTIAL™ can also be beneficial in improving the nutritional status of women prior to conception.

CONTRAINDICATIONS: Prenate® ESSENTIAL™ is contraindicated in patients with a known hypersensitivity to any of the ingredients.

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WARNING: Accidental overdose of iron-containing products is a leading cause of fatal poisoning in children under 6. Keep this product out of reach of children. In case of accidental overdose, call a doctor or poison control center immediately.

PRECAUTIONS: Folic acid alone is improper therapy in the treatment of pernicious anemia and other megaloblastic anemias where vitamin B₁₂ is deficient. Folic acid in doses above 1 mg daily may obscure pernicious anemia in that hematologic remission can occur while neurological manifestations progress.

ADVERSE REACTIONS: Anemia has been reported following both oral and parenteral administration of folic acid.

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PME-PI.1 Rev. 02/10

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Atlanta, Georgia USA 30338


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the proposed mechanism causing this finding in fetuses with abnormal karyotype.12 Although the incidence of chromosomal abnormalities is higher in fetuses with additional ultrasound markers of aneuploidy, echogenic bowel as an isolated finding still confers an increased risk (likelihood ratio, 6.7) for trisomy 21 above the maternal age-adjusted baseline risk.13 Cystic fibrosis also should be considered when echogenic bowel is diagnosed. It is thought that abnormal pancreatic enzyme secretion leads to thickened meconium, and subsequent meconium ileus is observed in 10% to 20% of newborns with cystic fibrosis.14 The estimated risk association between echogenic bowel and cystic fibrosis ranges from 0% to 13% in the literature.3,4,5,10,11,15 The finding of dilated loops of bowel in addition to echogenic bowel may increase this risk to as high as 17%.16 Finally, congenital infection also has been associated with isolated echogenic bowel. Direct damage to the fetal intestinal wall with subsequent paralytic ileus/perforation versus increased free peritoneal fluid secondary to hydrops may lead to the echogenic appearance of the bowel in cases of congenital infection. Cytomegalovirus (CMV) is the most commonly observed infection, but rubella, varicella, herpes, toxoplasmosis, and parvovirus also have been reported.1,3,10,11,17 Although the majority of studies quote a 3% to 4% incidence of congenital infection in fetuses with echogenic bowel, rates as high as 10% have been reported.3,10,11,17 In a series of 650 cases with primary CMV infection, 7 cases (1.2%) had isolated echogenic bowel as the sole ultrasound finding.18

What is the suggested evaluation for patients diagnosed with fetal echogenic bowel?
The finding of echogenic bowel on second-trimester ultrasound should prompt a detailed ultrasound examination in search for other anomalies. Strock et al demonstrated an increased incidence of additional anomalies, particularly renal and cardiac anomalies, in fetuses with echogenic bowel.4 If there is a normal 4-chamber view and the outflow tracts are visualized as normal, then a fetal echocardiogram is not necessary. However, if these views are abnormal or not obtainable, a fetal echocardiogram should be performed. Amniocentesis also should be offered to all patients irrespective of the presence or absence of associated anomalies, given that echogenic bowel is present as an isolated finding in 4% to 25% of fetuses with aneuploidy.3,8,9 Amniotic fluid should be sent for fetal karyotype as well as polymerase chain reaction for the most common congenital infections, such as CMV.

In patients who decline invasive testing, maternal IgG and IgM titers for CMV can be drawn. If these are suggestive of primary infection, then amniocentesis may be reconsidered. Without a history of exposure or other clinical risk factors, the yield of positive results for other congenital infections likely will be quite low and therefore should not be obtained routinely.

If unknown, parental cystic fibrosis carrier status also should be determined. Racial and ethnic limitations of current cystic fibrosis mutation screening panels should be taken into consideration when interpreting test results. If either parent is found to be a carrier, then genetic counseling can be performed to discuss the risks and benefits of invasive testing. Blood-stained amniotic fluid may suggest a prior intra-amniotic bleed that may be a contributing factor to the echogenic appearance of the fetal bowel.8 Although echogenic bowel has been reported in cases of homozygous alpha-thalassemia, testing for this disorder should be reserved only for high-risk populations, such as those of Southeast Asian or African descent.8 In the absence of prenatal diagnostic studies, pediatricians should be made aware of the antenatal finding of echogenic bowel at the time of delivery so that appropriate neonatal evaluation for congenital infection and cystic fibrosis can be performed. A suggested algorithm for the evaluation of fetuses with echogenic bowel is shown in Figure 2.

How should the patient be managed if no apparent etiology for the echogenic bowel can be determined?
The presence of echogenic bowel on fetal ultrasound, both when isolated and when present in combination with other anomalies, has been associated with intrauterine growth restriction (IUGR) and intrauterine fetal demise (IUFD).1,3,11,18,21–23 Serial evaluation of fetal growth by ultrasound assessment should be performed. Although Dicke et al observed either complete or partial resolution of echogenic bowel on serial ultrasound exams in normal fetuses, subsequent studies have demonstrated normal
fetal outcome even in the presence of persistent echogenic bowel.^[23,24] Persistent echogenicity of the fetal bowel should not be necessarily viewed as a marker for adverse outcome. Studies suggest most antenatal stillbirths occur at a previable or precarious gestational age range (22 to 24 weeks).[22,23] Although antenatal testing can be considered, its utility remains controversial in the management of isolated echogenic bowel unless fetal growth restriction or other indications develop.[25]

**REFERENCES**


