

Hyperechogenic Bowel

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During sonographic or ultrasound examination of the fetus, we can see the bowel or intestines within the fetal abdomen. Usually, the bowel is similar in level of "whiteness" in the ultrasound picture to surrounding structures, such as the liver and kidneys. In about 1 percent of pregnancies, the bowel appears to be much whiter than the surrounding structures, and this is called echogenic bowel or hyperechogenic bowel.

Hyperechogenic bowel is seen in about 1 percent of ultrasound examinations performed at the time of genetic sonography – between 15 and 22 weeks gestation. Hyperechogenic bowel is an important finding on ultrasound because, while over 90 percent of babies with this appearance to the bowel are perfectly normal, the sonographic finding flags the pregnancy for further evaluation.

Causes for hyperechogenic bowel include:

- The presence of blood within the amniotic fluid. For example, if Mom has had
 vaginal bleeding during the pregnancy, there will sometimes be bleeding into the
 amniotic space as well. The amniotic fluid may appear to have speckles of
 whiteness within it, and we can see speckles of white within the baby's stomach and
 a bright appearance to the bowel.
- **Chromosomal abnormalities** of the baby. Studies performed in the 1990s suggested that the risk for chromosome problems was as high as 3 to 7 percent, even if the remainder of fetal anatomic evaluation was completely normal. More recent data suggest that the risk for chromosome problems is in the **1 percent to 2 percent range.**
- **Viral infections** of the baby. The two most important of these are Cytomegalovirus infection and Parvovirus infection. Most women with CMV or Parvovirus infection will never be aware that they had the infection. We can screen for CMV and parvoviral infection by blood tests for mother (IgG and IgM testing). The presence of IgM suggests recent viral infection, and the diagnosis can be made by means of amniotic fluid testing obtained by amniocentesis. In the most recent studies, the chance of Cytomegalovirus and Parvovirus infection in pregnancies with hyperechogenic bowel has been in the **1 to 3 percent range**.

• **Cystic fibrosis.** Cystic fibrosis is a genetic condition that occurs in about 1 in 3,000 pregnancies. We can screen for cystic fibrosis by obtaining blood samples from the patient and the husband, and make the diagnosis by means of amniocentesis testing. In a recent series, the rate of cystic fibrosis in pregnancies with hyperechogenic bowel is **1 to 5 percent**.

There are other rare causes of hyperechogenic bowel. Homozygous alpha thalassemia (absence of genes for alpha hemoglobin) has been reported to cause hyperechogenic bowel, but these babies will virtually always show cardiac enlargement and enlargement of the umbilical vein.

The first step that we take when we identify hyperechogenic bowel (bowel that appears to be as bright as bone) is to complete a comprehensive evaluation of the baby – a "genetic sonogram." You will find a description of the <u>genetic sonogram</u> elsewhere on our website. If other issues are detected at the time of genetic sonography, then management and recommendations, of course, will be individualized.

If the sonogram is normal apart from hyperechogenic bowel, then typical recommendations are:

- 1. Genetic counseling.
- 2. Cystic fibrosis screening or testing.
- 3. Screening or testing for Cytomegalovirus and parvoviral infection, and;
- 4. Consideration of amniocentesis for chromosome testing, and direct testing for cmv, parvovirus, and cystic fibrosis.

Once these results are back then we recommend:

- 1. More or less monthly scans to assess fetal growth, because the finding of echogenic bowel raises the chance that the baby develops growth problems before birth.
- 2. A plan for careful evaluation of the baby after delivery by a pediatrician who is aware of the findings in utero and knows to check the baby carefully for signs of intestinal problems.

The finding of echogenic bowel on ultrasound is frightening and concerning for families. It is important to understand that over 90 percent of babies with echogenic bowel will prove to be completely normal, and that in a substantial proportion of the remainder, the finding of echogenic bowel will lead to a diagnosis, such as parvoviral infection,

CMV infection, or cystic fibrosis, which can either be treated before birth, or for which the in utero diagnosis will be important in planning treatment after the baby is born.