Fetal Echogenic Bowel

Recommendations of Practice

Background

- Assessment of the fetal bowel is a routine component of the second trimester screening ultrasound. Echogenic bowel (EB) is identified as an increased brightness (echogenicity) of the fetal bowel and can be focal or multifocal. In isolation, EB is often a benign finding and the majority of fetuses are normal. However, EB can be associated with anomalies (i.e. genetic, anatomic, infectious, growth restriction) and further assessment is warranted as this carries an increased risk of morbidity and mortality.

Objective

- To guide accurate diagnosis, investigations, and management of fetuses with echogenic bowel.

Definition

- Ultrasound finding of fetal bowel echogenicity at least equal to or greater than surrounding bone echogenicity.

Differential Diagnosis

- The majority of fetuses with isolated fetal EB have a normal outcome (>90%). In cases with an underlying pathology, additional ultrasound findings are usually identified and EB is not isolated. In these cases, the prognosis will depend upon the diagnosed condition.
    - Idiopathic (>80%)
        - In cases where EB is isolated and no other ultrasound, genetic, or infectious abnormalities are identified, the outcome is generally normal.
    - Chromosomal (3-11%)
        - The most common chromosomal anomaly detected in fetuses with EB is Trisomy 21. Additional anomalies include Trisomy 18 and 13, Turners syndrome and other unbalanced translocations
    - Cystic fibrosis (~2-3%)
    - Congenital viral infection (1-6%)
        - Primarily seen with cytomegalovirus infection but also reported with toxoplasmosis and Parvo virus infection
    - Fetal growth restriction (10-12%)
        - EB seen with early FGR has been associated with pre-eclampsia and HELLP syndrome later on in gestation
    - Fetal ingested blood

        - This can occur in the context of intra-amniotic haemorrhage. The EB tends to resolve with time with a normal outcome
    - Bowel malformation (6%)
        - EB has been seen in the context of fetal bowel obstruction, atresia, and perforation.

Maternal history

- Obstetric or family history of aneuploidy, cystic fibrosis, congenital anomalies
- Illnesses during pregnancy
- Vaginal bleeding during pregnancy or early ultrasound showing subchorionic bleed
- A priori risk for aneuploidy.

Ultrasound diagnosis

- 2nd trimester ultrasound
- Use transducer frequency ≤ 5 MHz
- Compare echogenicity of bowel to nearby fetal bone (as bright as bone)
- Turn down gain to better assess level of echogenicity
- Determine if focal or multifocal
- Assess for markers of aneuploidy, structural anomalies, bowel anomalies, markers for infection, subchorionic haematoma, growth restriction.

Investigation and Management

- Counselling for additional testing should take into consideration the ultrasound findings, a priori risk of aneuploidy, obstetric and family history, and parental wishes.
    - Amniocentesis (usually reserved for those cases with additional US findings)
        - Genetic testing for aneuploidy (FISH and microarray); consider cystic fibrosis testing depending on population incidence, family history, and parental carrier status (see below)
        - Infection testing (PCR for CMV, Toxoplasmosis, Parvovirus)
    - NIPT
        - NIPT can be offered to those women who decline amniocentesis. However, these patients must be counselled that cell-free DNA is a screening test and if positive, an amniocentesis would be recommended for diagnostic confirmation
    - Maternal serology
        - IgM and IgG for CMV, toxoplasmosis, and Parvovirus
    - Parental testing for cystic fibrosis carrier status
        - Detects 80% of common mutations
        - Not all mutations can be identified and this does not exclude the possibility
        - If both parents are carriers fetal diagnostic testing maybe offered, genetic counselling is strongly recommended.
    - Serial scanning
        - Assess resolution or persistence of EB
        - Identification of anomalies not seen previously
        - Monitoring fetal growth in the third trimester.

Prognosis

- In cases of isolated EB, the prognosis is favourable for a normal outcome (>90%). In non-isolated EB, outcome will depend on other associated findings and diagnosis.

This Recommendation of Practice was updated in March 2023 by Dr Kristy Wolff with input from members of Wāhi Rua NZMFM Network.

The most up to date version of this Recommendation of Practice can be found on Healthpoint Wāhi Rua: New Zealand Maternal Fetal Medicine Network (NZMFM) webpages: https://www.healthpoint.co.nz/public/wahi-rua-new-zealand-maternal-fetal-medicine/

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