



# Etiologies and Outcomes of Fetal diagnosis of echogenic bowel

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## INTRODUCTION

Soft markers are variants of normal findings on ultrasound but important as indicate increased risk for fetal aneuploidy. Common soft markers include: nuchal fold thickness, ventriculomegaly, cardiogenic focus, choroid plexus cysts, and echogenic bowel (EB). EB (Figure 1) is associated with increased risk in fetal Aneuploidy (increases risk of T21 6x, increases risk for T13 and T18), cystic fibrosis, congenital infections (CMV), intra-amniotic bleeding, congenital malformations of bowel, other perinatal complications (IUGR). However, little is known about underlying etiologies of echogenic bowel, resulting in little to no follow up for patients.

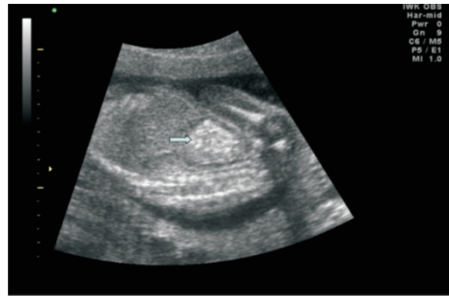


Figure 1. Fetal echogenic bowel on ultrasound.

## AIMS

1. To determine the prevalence of echogenic bowel in MB and frequency of unexplained.
2. To determine the most common etiologies underlying echogenic bowel and associated fetal outcomes

## METHODS

This was a historical cohort study looking at cases from Jan 1, 2012- Dec 31, 2018. All fetuses with EB were identified using a regional, prenatal genetics

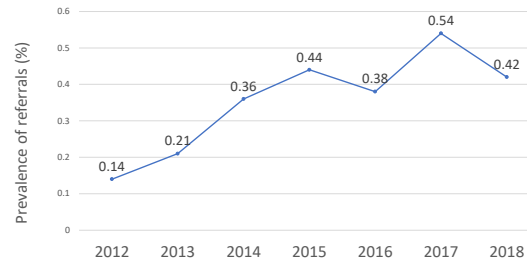


Figure 2. Temporal trends in echogenic bowel referrals in Manitoba.

clinical database. Cases of EB were designated as 'Isolated' or 'Complex' depending on the coexistence of additional ultrasound or analyte abnormalities. Stored consult letters and ultrasound reports reviewed by independent research personnel. A standard data collection sheet was used and included maternal demographics, fetal US and testing results. Descriptive and inferential statistics were used to compare findings between groups. Prevalence of fetal EB was calculated using the total number of livebirths per year as the denominator.

## RESULTS

Of the 252 EB referrals, 232 cases were eligible and included in the final cohort. The prevalence of referrals for EB increased significantly over the study period (0.14% to 0.42%;  $p=0.038$ ) (Figure 2). 77.3% of cases were referred with isolated EB while the remaining were found to be complex cases (Figure 3). 11.2% of all cases were found to have underlying diagnoses while the majority remained unexplained. There was no difference in maternal age, parity, or residence between cases of isolated versus complex EB. Of those with an underlying diagnosis, anomalies and cystic fibrosis were the most common etiologies in the Isolated EB group: in the Complex EB group, the most common diagnosis was genetic (aneuploidy or syndromic) but no cases of cystic fibrosis.

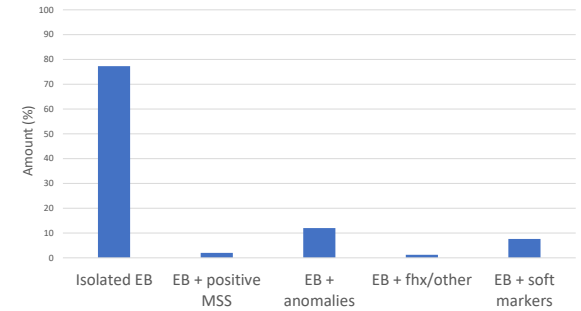


Figure 3. Proportion of cases of isolated versus complicated echogenic bowel (EB).

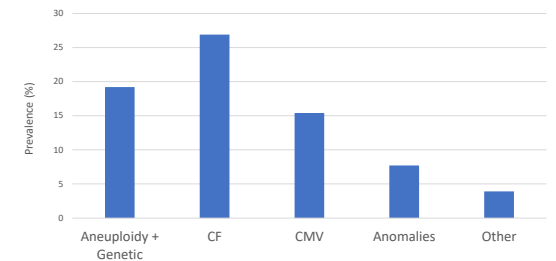


Figure 4. Explained Echogenic Bowel Underlying Etiologies

## CONCLUSION

The majority of cases of EB remain unexplained, and very few have T21. Of those with an underlying diagnosis, cystic fibrosis and anomalies were the most common etiologies identified in the isolated EB group whereas other genetic causes (syndromes or aneuploidy) were predominant causes in the complex EB group. Better understanding of the underlying causes of EB will improve counseling of patients and families, and direct need for additional testing and follow-up.