

MFM INTERESTING CASE E-MAIL BROADCAST

Dear Colleague,

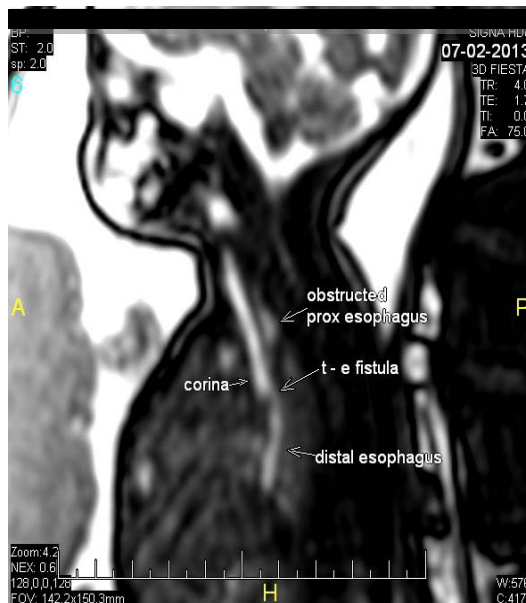
Welcome back to MFM Interesting Case presentations. In the year since prior broadcast I have performed 380 fetal MRI studies for the tri-state MFM community.

The accompanying two cases highlight unique attributes of fetal MR in problem solving evaluation for non CNS pathologies.

Case 1 – Esophageal atresia with tracheo-esophageal fistula

This is a 32-week gestation whose outside ultrasound studies demonstrated an absent stomach with a known Down syndrome karyotype. Outside ultrasound studies specifically sought, but could not identify, a dilated proximal esophageal “pouch” sign.

In the MR evaluation for esophageal atresia special pulse sequences are necessary. The 2 mm T2 volume acquisition sequence is very fast. This sequence is repeatedly performed over ten minutes to capture a fetal swallowing sequence. The submitted images demonstrate the dilated esophagus proximal to the level of atresia with the distal esophagus communicating to the carina at the level of the tracheoesophageal fistula.



The patient was delivered at a tertiary care center. Findings were confirmed at surgery.

Teaching Point: MR is capable of the direct diagnosis of esophageal atresia and tracheoesophageal fistula employing rapid T2 volume pulse sequence acquisition.

Discussion: Consideration should be given to fetal MR in the evaluation for possible esophageal atresia with or without TE fistula under several clinical scenarios including – but not limited to - unexplained polyhydramnios, fetuses demonstrating stigmata that might be part of a VACTERL association and in fetuses with other gastrointestinal pathologies.

Antenatal diagnosis of esophageal atresia will necessitate either delivery at, or transfer to, a tertiary care center for surgical reconstruction. Antenatal diagnosis of esophageal atresia should prompt recommendation of karyotyping given the 6 to 10% association with chromosomal abnormalities. Esophageal atresia and/or tracheoesophageal fistula is found in 1% of infants with T21, 25% of infants with T18. Other chromosomal abnormalities that are associated with esophageal atresia and TE fistula include deletions of bands 17q 22-23, 13q 32 and 22q 11.

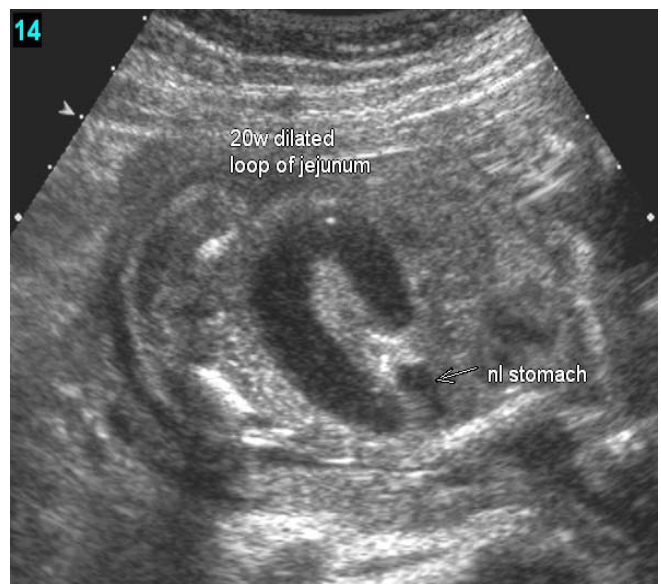
Fetal MR offers the opportunity for second trimester diagnosis of esophageal atresia prior to the third trimester onset of polyhydramnios.

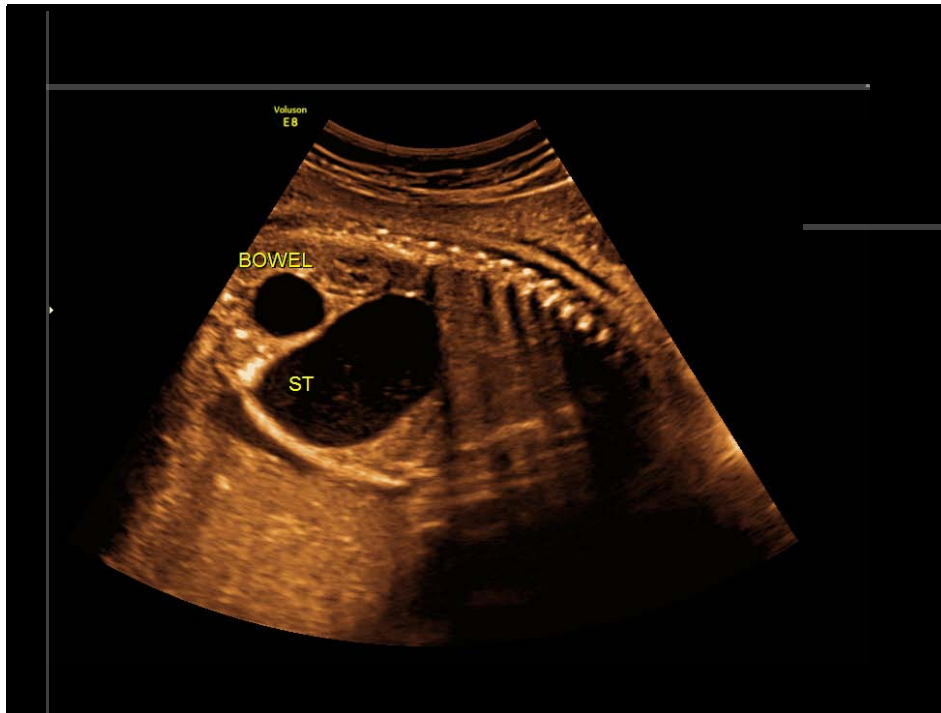
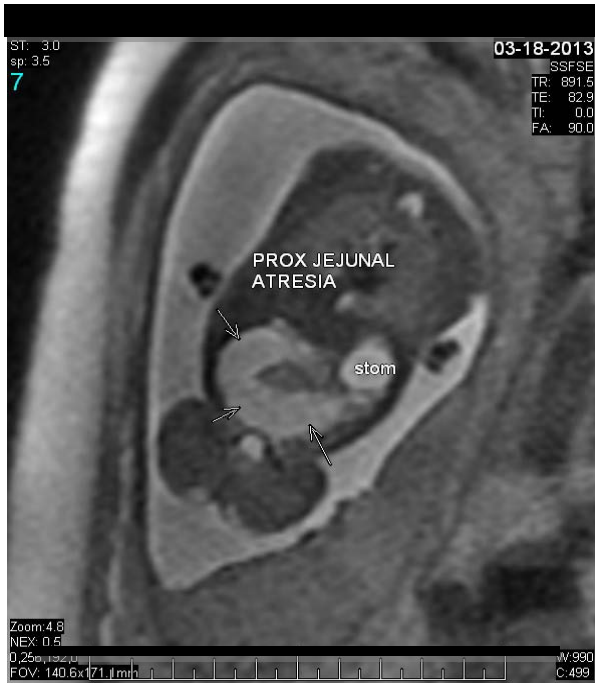
Case 2 – Multiple intestinal atresias in association with combined immunodeficiency syndrome

History: 31-year-old IVF pregnancy whose 20-week detailed ultrasound demonstrated a solitary c-shaped dilated loop of upper abdominal intestine, extensive intraabdominal/pelvic echogenic foci, and normal caliber stomach. No signs of congenital heart disease. Karyotype, cystic fibrosis, and infectious studies were normal.

Fetal MR was performed with the imaging goals to establish small bowel versus colonic organ of origin of the dilated intestinal loop and to evaluate for any associated gastrointestinal pathologies such as esophageal atresia, TE fistula, and anal atresia.

MR demonstrated the dilated loop of bowel to be of jejunal origin. MR demonstrated no signs of esophageal atresia or TE fistula and demonstrated a normal caliber rectum extending to the anal region excluding all but the most caudal patterns of imperforate anus/anal atresia.





The patient's care was transferred to a tertiary care center. The appearance of the bowel was unchanged on follow up ultrasound studies up until 32 weeks at which time the stomach was noted to be markedly dilated with onset of polyhydramnios. No additional dilated loops of bowel were identified distal to the obstructed proximal jejunal loop.

Surgical findings – At the time of surgery multiple sites of atresia were identified including the pylorus, small and large bowel. The small bowel was extensively filled with significant amounts of inspissated material. Seven sites of bowel resection with end-to-end anastomoses was performed.

Neonatal medical evaluation diagnosed combined immunodeficiency disorder. At two-months of age the patient remains in the ICU with no functioning bowel. The ultimate prognosis is quite guarded. Management issues being contemplated include bone marrow transplantation and possibly intestinal transplantation.

Teaching Point: When intestinal pathologies are suspect on ultrasound, the value of MR is to assign organ of origin – small bowel versus larger bowel, and to evaluate for associated gastrointestinal pathologies that are difficult to diagnose on ultrasound including esophageal atresia +/- TE fistula and anal atresia.

Discussion: Pyloric atresia causing gastric outlet obstruction in combination with jejunal atresia is unusual. Whole exomic sequencing has identified mutations of the TTC 7A gene in association with multiple intestinal atresias and immune deficiency syndrome. Immunodeficiency syndromes should be considered in the differential diagnosis when multiple sites of intestinal obstruction are diagnosed antenatally. (J Pediatric Surg. 1998. May;33 (5):794-7).

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