Prenatal Diagnosis of Esophageal Atresia Using Sonography and Magnetic Resonance Imaging

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Background: The diagnosis of esophageal atresia may be suspected on prenatal ultrasound scan in fetuses with a small or absent stomach or unexplained polyhydramnios. However, these findings are thought to have a low positive predictive value and clinical decisions affecting timing or site of delivery may be made erroneously. The authors evaluated the accuracy of fetal sonography followed by magnetic resonance imaging (MRI) for the diagnosis of this lesion.

Methods: Fetuses considered to be at risk for esophageal atresia based on detailed obstetric sonography underwent fetal MRI using a single-shot rapid-acquisition technique, and the T_2-weighted images were evaluated prospectively. Scans were considered to be positive if the proximal esophagus was dilated, and the distal esophagus was not seen and negative if the esophagus was visualized throughout its length.

Results: Ten fetuses underwent MRI scanning. All had a small or absent stomach bubble with unexplained polyhydramnios. Four scans were considered to be negative for esophageal atresia; all 4 were found to have a normal esophagus after delivery. Six scans were considered to be positive; 5 had esophageal atresia (2 with tracheoesophageal fistula and 3 without), and one had a neurologic syndrome with a normal esophagus.

Conclusions: Magnetic resonance imaging appears to be accurate for establishing or ruling out a prenatal diagnosis of esophageal atresia, and should be considered in fetuses who are at high risk based on ultrasound findings.

INDEX WORDS: Prenatal diagnosis, prenatal ultrasound scan, polyhydramnios, magnetic resonance imaging.

UNLIKE many other anomalies commonly seen by pediatric surgeons, esophageal atresia rarely is diagnosed prenatally. In retrospective studies, only one third of affected infants are identified successfully by prenatal ultrasound scan.[1-3] The findings that suggest the possibility of esophageal atresia are small or absent stomach bubble, unexplained polyhydramnios, or chromosomal abnormalities such as trisomy 18, which are known to be associated with esophageal atresia. However, the predictive value of these ultrasound findings also is very low, with 56% to 83% of the infants who are suspected of having esophageal atresia sonographically ultimately having a normal esophagus seen when the child is born.[3,4]

Magnetic resonance imaging (MRI) has been used increasingly to image the fetus.[5] Studies have shown the utility of MRI for visualization of the central nervous system and urinary tract and for estimation of fetal weight, with no significant fetal or maternal morbidity.[6-8] MRI also has been used to image a number of pediatric surgical conditions in utero.[9,10] The goal of the current study was to evaluate the accuracy of MRI for the diagnosis of esophageal atresia in the fetus with a suspicious prenatal sonogram.

MATERIALS AND METHODS

A prospective study was performed between January 1997 and July 2000 at Washington University and the University of Toronto. Fetuses were identified as high risk for esophageal atresia based on ultrasound identification of a small or absent stomach with unexplained polyhydramnios. The mother was offered the opportunity to undergo fetal MRI, and appropriate informed consent was obtained in all cases.

All MR imaging was performed with a 1.5 T superconducting magnet (Signa CV/i, Signa Horizon LX; GE Medical Systems, Milwaukee, WI) using phased-array surface coils. T_2-weighted images were obtained using a single-shot rapid-acquisition technique. Axial, coronal, and sagittal planes were performed relative to the fetus. A variable bandwidth was utilized. The slice thickness ranged from 4 to 6 mm with an interslice gap of 0 to 1 mm. The field of view was optimized for each patient.

Each scan was reviewed prospectively by a pediatric surgeon and a radiologist, and was called either negative or positive for esophageal atresia. Scans were considered negative if the entire esophagus could be visualized and positive if the esophagus was not seen in the midchest. All infants were delivered at a perinatal center and evaluated by the neonatal and surgical teams. Evaluation included passage of a nasogastric tube, complete physical examination, and radiologic studies if necessary. A postnatal diagnosis of either esophageal atresia or normal esophagus was made.

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Table 1. Summary of Patient Data

Patient No.	Prenatal Sonographic Findings		MIR Diagnosis	Postnatal Diagnosis	Other Diagnoses
	Stomach	"Pouch" Sign			
1	Small	No	No EA	Normal esophagus	Trisomy 18
2	Small	No	No EA	Normal esophagus	None
3	Small	No	No EA	Normal esophagus	None
4	Small	No	No EA	Normal esophagus	Congenital heart disease, Dandy-Walker variant, CHARGE association
5	Small	No	EA	EA/TEF	Hydronephrosis
6	Small	No	EA	Normal esophagus	Rubinstein-Taybi syndrome
7	Absent	Yes	EA	Pure EA	Microcephaly, VSD
8	Absent	No	EA	Pure EA	None
9	Absent	No	EA	EA/TEF	Horseshoe lung Esophageal bronchus
10	Absent	No	EA	Pure EA	None

Abbreviations: EA, esophageal atresia; TEF, tracheoesophageal fistula; VSA, ventricular septal defect.

The data were analyzed using standard 2 × 2 tables to calculate sensitivity, specificity, and positive predictive value.

RESULTS

Ten fetuses underwent MRI scanning between 23 and 34 weeks of gestation (median, 31 weeks). In these fetuses, a small or absent stomach was noted sonographically between 17 and 33 weeks of gestation (median, 23 weeks). The cases are summarized in Table 1. Examples of both negative and positive scans are shown in Figs 1 and 2, respectively.

Four fetuses had scans that were considered to be negative for esophageal atresia. All 4 were found not to have esophageal atresia postnatally. Of these, 1 was known to have trisomy 18, 1 had congenital heart disease and a Dandy-Walker variant, and 2 were normal newborns. Six fetuses had scans that were considered to be positive. Of these, 5 were found to have esophageal atresia. Three had pure atresia, and 2 had an associated tracheoesophageal fistula. One of these had a complex group of anomalies including right upper lobe tracheal bronchus, right lower lobe esophageal bronchus, horseshoe lung, and annular pancreas. In this fetus, the prenatal MRI showed an abnormal right upper lobe in addition to the findings in the esophagus (Fig 3). One fetus had a positive scan and was not found to have esophageal atresia postnatally, representing the only false-positive result in the study. This child ultimately was felt to have Rubenstein-Taybi syndrome, which included neurologic impairment with poor swallowing as well as severe gastroesophageal reflux.

Overall, the sensitivity of MRI in this study was 100% (ie, all infants with esophageal atresia had a positive scan), the specificity was 80% (ie, 4 of 5 infants without esophageal atresia had a negative scan), the positive predictive value was 83% (ie, 5 of the 6 positive scans correctly predicted the presence of esophageal atresia), and the negative predictive value was 100% (ie, all 4 negative scans correctly predicted a normal esophagus).

We also evaluated the accuracy of prenatal sonography in these patients. The overall positive predictive value of ultrasound scan was only 60% (ie, 6 of 10

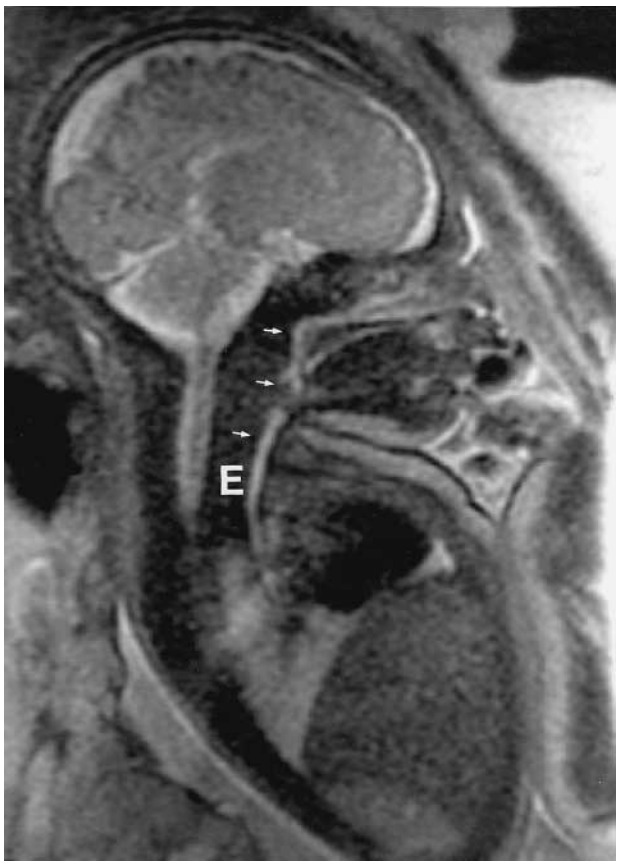

Fig 1. Fetal MRI image (sagittal view) that is negative for esophageal atresia. Note the normal-sized hypopharynx and proximal esophagus (arrows), and the normal intrathoracic esophagus (E).

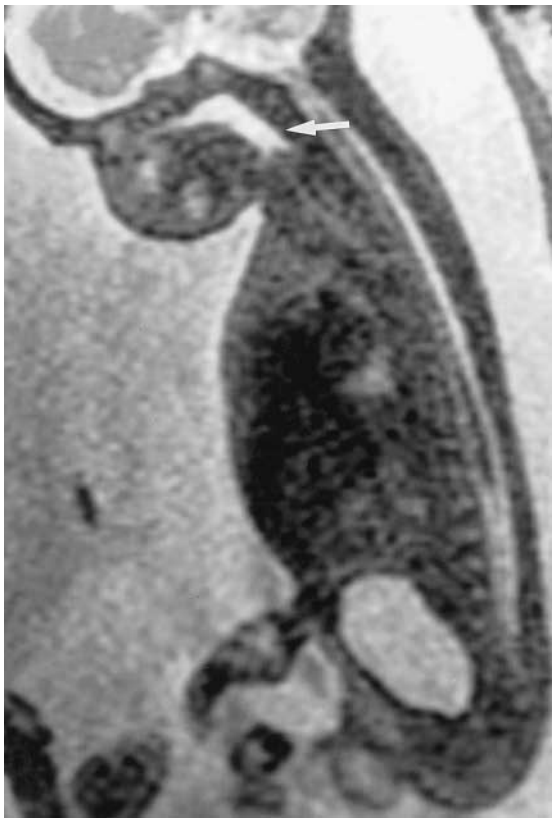

Fig 2. Fetal MRI image (sagittal view) that is positive for esophageal atresia. Note that the hypopharynx and proximal esophagus are dilated (arrow) and that there is no discernable intrathoracic esophagus.

correctly predicted the presence of esophageal atresia) and was only 33% in those with a small stomach. Three of the fetuses had a normal-size stomach on some sonograms and a small stomach on others (patients 2, 5, and 6); 2 of these had a normal esophagus at delivery, and 1 had esophageal atresia with tracheoesophageal fistula. Importantly, all 4 fetuses with an absent stomach had esophageal atresia. Only 1 fetus was found to have a positive "pouch" sign on ultrasound scan, and this infant had pure esophageal atresia at delivery.

DISCUSSION

The prenatal diagnosis of esophageal atresia is known to be inaccurate. Several studies have documented that the sensitivity of sonography is only 24% to 30% for the prenatal detection of this anomaly.[1-3] Because many fetuses with esophageal atresia may not have significant polyhydramnios or sonographic abnormalities, there currently is no way to improve our sensitivity in detecting this anomaly.

This study was designed to address the clinical scenario that occurs when a sonographic finding of a small or absent stomach associated with polyhydramnios raises the suspicion of esophageal atresia. In most of these cases, the timing or location of delivery, as well as the direction and content of prenatal counseling, may be altered significantly because of the sonographic findings.[11] Experience would suggest that despite the prenatal findings, many of these children are found to have a normal esophagus postnatally. Our data suggest that fetal MRI is an accurate and noninvasive way to clarify the diagnosis in fetuses at high risk based on the sonographic findings. If the MRI is normal, and there are no other abnormalities that would dictate delivery at a perinatal center, the family can be counseled appropriately, and delivery at a local hospital can be recommended. If abnormal, the MRI leads to appropriate preparation of both the family and the health care team.

Recently, several investigators have suggested that identification of a fluid-filled pouch in the neck or mediastinum may improve the predictive value of the ultrasound scan.[12,13] However, this was only seen in 1 fetus in our series, and there were several fetuses without this finding who had positive MRI scans and esophageal atresia at the time of delivery. We found that complete absence of the stomach bubble on repeated sonograms was a more accurate predictor than identification of a fluid filled pouch. It is possible that complete absence of the stomach bubble may be predictive enough to eliminate the need for MRI, but a larger experience will be necessary to reliably draw this conclusion.

The safety of fetal MRI has been studied in both animal models and in humans. Several early studies in rodents suggested that fetal MRI may interfere with development of the eyes and face, and may impair fetal growth slightly.[14,15] However, these results often were contradictory, and a number of clinical studies have failed to show any evidence of harm to human fetuses.[16-18]

In this small group of patients, the sensitivity of MRI

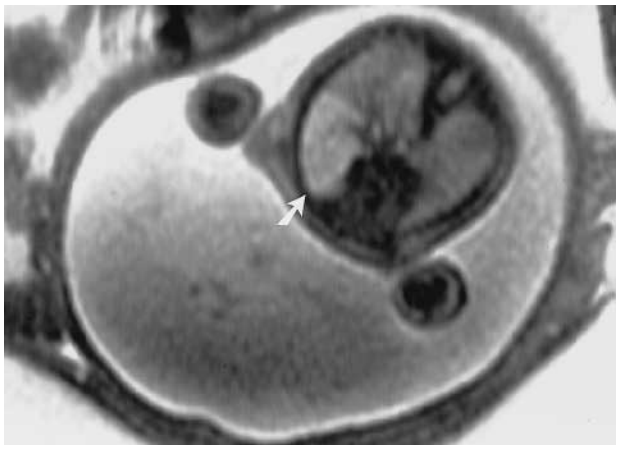

Fig 3. Transverse fetal MRI image shows an abnormal right upper lobe of the lung (arrow) in patient 9.

was 100%. However, there was one false-positive in a patient whose small stomach and polyhydramnios presumably were caused by poor swallowing from a neurologic syndrome. Even though this patient did not have esophageal atresia, the delivery at a perinatal center that resulted from the positive MRI was beneficial to the patient. Although we did not have any in this series, a false-negative scan would be more concerning, because it might lead to delivery of a child with esophageal atresia at a community hospital. A larger experience with this technique will be necessary to confirm an acceptably low false-negative rate.

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