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Esophageal atresia and tracheoesophageal fistula: prenatal sonographic manifestation from early to late pregnancy

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KEYWORDS: absent stomach; esophageal atresia; esophageal pouch; polyhydramnios; small stomach; tracheoesophageal fistula

CONTRIBUTION

What are the novel findings of this work?
Our study is the first to elucidate the sonographic manifestation of esophageal atresia and/or tracheoesophageal fistula (EA/TEF) as early as 14 weeks' gestation. It is also the first to quantify the yield of anomaly scans performed at different stages of pregnancy. We found that an early anomaly scan can detect indirect signs of EA/TEF as early as 15 weeks, but in only a minority of cases.

What are the clinical implications of this work?
Anomaly scans in their current form perform poorly in screening for and diagnosing EA/TEF. In approximately 30% of cases, sonographic signs are absent. We suggest different strategies to improve diagnostic performance for EA/TEF.

ABSTRACT

Objective *Esophageal atresia and/or tracheoesophageal fistula (EA/TEF) remains one of the most frequently missed congenital anomalies prenatally. The aim of our study was to elucidate the sonographic manifestation of EA/TEF throughout pregnancy.*

Methods *This was a retrospective study of data obtained from a tertiary center over a 12-year period. The prenatal ultrasound scans of fetuses with EA/TEF were assessed to determine the presence and timing of detection of three principal signs: small/absent stomach and worsening polyhydramnios, both of which were considered as 'suspected' EA/TEF, and esophageal pouch, which was considered as 'detected' EA/TEF. We assessed the yield of the early (14–16 weeks' gestation), routine mid-trimester (19–26 weeks) and third-trimester (≥ 27 weeks) anomaly scans in the prenatal diagnosis of EA/TEF.*

Results *Seventy-five cases of EA/TEF with available ultrasound images were included in the study. A small/absent stomach was detected on the early anomaly scan in 3.6% of fetuses scanned, without a definitive diagnosis. On the mid-trimester scan, 19.4% of scanned cases were suspected and 4.3% were detected. On the third-trimester anomaly scan, 43.9% of scanned cases were suspected and 33.9% were detected. An additional case with an esophageal pouch was detected on magnetic resonance imaging (MRI) in the mid-trimester and a further two were detected on MRI in the third trimester. In total, 44.0% of cases of EA/TEF in our cohort were suspected, 33.3% were detected and 10.7% were suspected but, eventually, not detected prenatally.*

Conclusions *Prenatal diagnosis of EA/TEF on ultrasound is not feasible before the late second trimester. A small/absent stomach may be visualized as early as 15 weeks' gestation. Polyhydramnios does not develop before the mid-trimester. An esophageal pouch can be detected as early as 22 weeks on a targeted scan in suspected cases. The detection rates of all three signs increase with advancing pregnancy, peaking in the third trimester. The early and mid-trimester anomaly scans perform poorly as a screening and diagnostic test for EA/TEF.* © *2020 International Society of Ultrasound in Obstetrics and Gynecology.*

INTRODUCTION

Esophageal atresia and/or tracheoesophageal fistula (EA/TEF) occurs in one in 2500–3800 live births[1–4].

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Despite improvements in ultrasound technology, EA/TEF remains one of the most frequently missed diagnoses prenatally[2,5–7]. The challenges in the prenatal diagnosis of EA/TEF are numerous. Polyhydramnios and a small/absent stomach bubble are neither sensitive nor specific signs and they do not appear consistently on prenatal scans[2,5,7–9]. The appearance of these signs is probably influenced by the type of EA/TEF[10] and the gestational age at the time of the ultrasound scan. Visualization of the whole length of the esophagus, when intact, or demonstration of an esophageal pouch on ultrasound is time consuming and requires experience[11–13]. Furthermore, scanning the esophagus on routine anomaly scans is not mandatory in international guidelines[14].

The aim of our study was to elucidate the manifestation of the three sonographic signs of EA/TEF (a small/absent stomach, polyhydramnios and esophageal pouch) from the early anomaly scan (14–16 weeks' gestation, as practiced in Israel) to the third-trimester anomaly scan.

METHODS

This was a retrospective study of data of all pregnancies affected by fetal EA/TEF, either isolated or non-isolated, obtained at our medical center between 2006 and 2019. The prenatal ultrasound images of these cases were assessed for the presence and timing of the appearance of the three principal signs of EA/TEF: polyhydramnios (maximal vertical pocket (MVP) ≥ 8 cm or amniotic fluid index (AFI) ≥ 24 cm) and small/absent stomach (Figure 1a,b), which were considered suspicious signs, and esophageal pouch (Figure 1c), which was considered diagnostic. The severity of polyhydramnios was recorded, with severe polyhydramnios defined as a MVP ≥ 16 cm or an AFI ≥ 35 cm. Postnatally, all liveborn cases were diagnosed on chest X-ray with contrast medium. In the few cases of fetal demise or termination of pregnancy, postmortem diagnosis was unavailable, and the diagnosis of EA/TEF was based on prenatal findings.

We reviewed medical records including ultrasound reports, images and videoclips, magnetic resonance imaging (MRI) reports, genetic and laboratory work-up, obstetric clinic visits, labor-ward reports and neonatal intensive care unit reports.

In Israel, a routine mid-trimester anomaly scan (19–26 weeks) is recommended and subsidized for all women. Women can choose to have an additional early second-trimester anomaly scan (14–16 weeks) at partial subsidization. A third-trimester scan (at or after 27 weeks) is usually performed in cases that are at risk for anomalies or abnormal growth, or on maternal request. We assessed the contribution of each of the three anomaly scans (the early, the routine mid-trimester and the third-trimester scans) to the rate of suspicion (small/absent stomach or worsening polyhydramnios) and the rate of detection (pouch sign, on either ultrasound or MRI) of EA/TEF. MRI was performed at the discretion of the sonographer in suspected cases in which ultrasound failed to determine or refute the diagnosis.

The study protocol was approved by the Institutional Ethical Committee at Chaim Sheba Medical Center (approval number 5238-18-SMC).

Statistical analysis

Normality of the data was tested using the Shapiro–Wilk or Kolmogorov–Smirnov test. Data are presented as median (interquartile range), median (range), *n* (%) or *n*/*N* (%). Comparisons between unrelated variables were conducted using Student's *t*-test or the Mann–Whitney *U*-test, as appropriate, and the χ^2 test or Fisher's exact test was used for comparisons between categorical variables. Stepwise logistic regression analysis was performed for multivariate categorical analysis. Statistical analysis was conducted using SPSS v.23 (IBM Corporation Inc., Armonk, NY, USA); $P < 0.05$ was considered to indicate statistical significance.

RESULTS

Seventy-six cases of EA/TEF were included in the study. One of the cases did not undergo ultrasound scans during pregnancy and was therefore included only in the analysis of maternal demographics and peri/postnatal data. Of

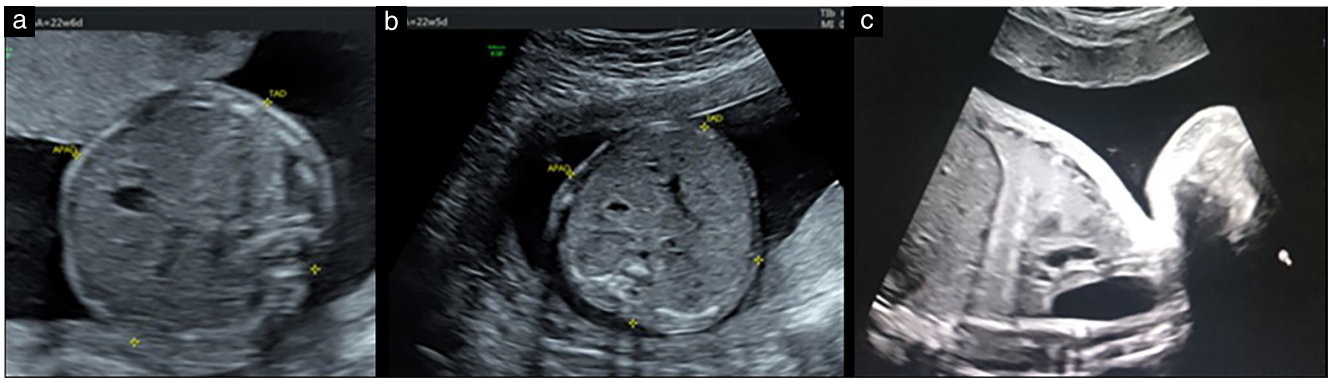

Figure 1 Sonographic signs of esophageal atresia and/or tracheoesophageal fistula: (a) absent stomach; (b) small stomach; (c) esophageal pouch.

the 76 cases, 71 were liveborn, two were stillborn and three were terminated. Among the 75 cases that were scanned throughout pregnancy, 55 had an early anomaly scan, 69 had a routine mid-trimester scan and 56 had a third-trimester scan, either routine ($n = 4$), targeted for various conditions other than EA/TEF ($n = 19$) or owing to suspicion of EA/TEF ($n = 33$). Once a case was suspected on a scan, it was omitted from further analysis of suspicion on subsequent scans. The background details of the patients are presented in Table 1.

Figure 2 summarizes the number of cases undergoing each anomaly scan (early, routine mid-trimester and third-trimester) and how many cases were suspected (small/absent stomach and/or worsening polyhydramnios) and detected by demonstration of an esophageal pouch at each scan.

Overall, 33 of 75 (44.0%) cases were suspected prenatally on ultrasound to have EA/TEF. Of these, 25 cases were diagnosed prenatally with EA/TEF by detection of an esophageal pouch on ultrasound and/or MRI (33.3% overall prenatal detection rate) and the remaining eight (10.7%) suspected patients were not diagnosed (two were assumed to have EA/TEF without pouch detection, three were falsely ruled out as 'normal' and three declined further tests).

The rate of EA/TEF suspicion and detection and the rate of appearance of each indirect sign on each of the three anomaly scans are compared in Table 2. Clear and statistically significant increase in the rates of EA/TEF suspicion, detection and appearance of each indirect sign were apparent with advancing gestational age at the time of the anomaly scan.

Table 1 Demographic characteristics of study group of 76 pregnancies with fetal esophageal atresia and/or tracheoesophageal fistula

Characteristic	*Value*
Maternal age (years)	32 (28–35)
Nulliparous	28 (36.8)
Prepregnancy BMI (kg/m^2)	24 (21–29)
Prepregnancy BMI > 30 kg/m^2	15/57 (26.3)
IVF pregnancy	14 (18.4)
Multiple pregnancy	17 (22.4)

Data are given as median (interquartile range), *n* (%) or *n*/*N* (%). BMI, body mass index; IVF, *in-vitro* fertilization.

Contribution of early anomaly scan to EA/TEF suspicion and detection

In 2/55 cases scanned for anomalies at 14–16 weeks, there was a persistent small/absent stomach, yielding a 3.6% EA/TEF suspicion rate for the early anomaly scan. Both cases had an esophageal pouch detected later on in pregnancy, at 22.6 and 28.4 weeks' gestation, respectively. Overall, 7.4% (2/27) of patients diagnosed with a small/absent stomach at any of the three scans were identified before 16 weeks. None of the cases was diagnosed with an esophageal pouch or displayed polyhydramnios at the time of the early anomaly scan.

Contribution of routine mid-trimester scan to EA/TEF suspicion and detection

There were 67 cases that had a routine mid-trimester anomaly scan and were not suspected previously for EA/TEF (Figure 2). Of these, 13 (19.4%) cases were reported to have a small/absent stomach and/or worsening polyhydramnios on this scan, raising suspicion for

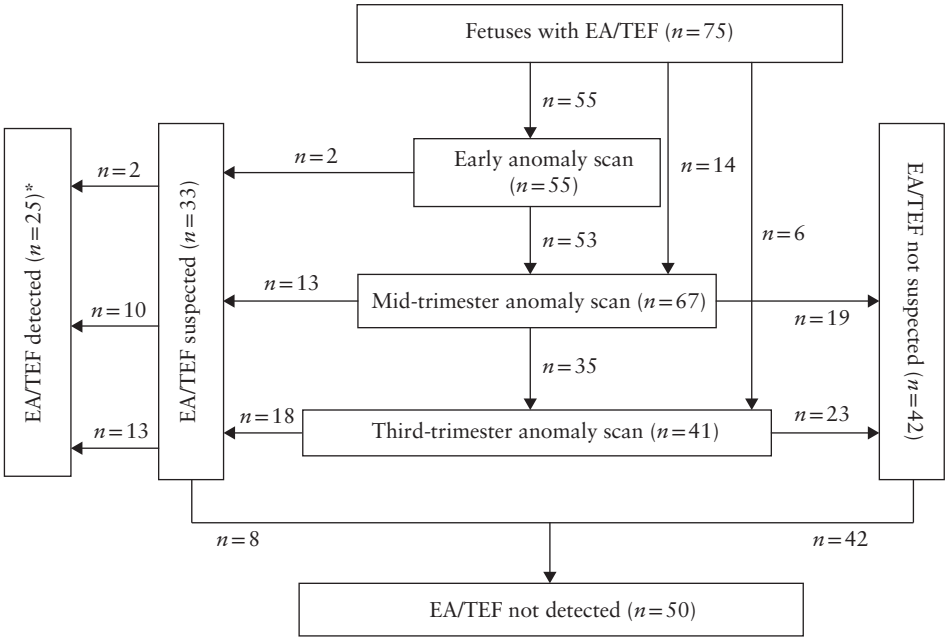

Figure 2 Flowchart summarizing prenatal evaluation of 75 fetuses with esophageal atresia and/or tracheoesophageal fistula (EA/TEF). *Three of the 25 detected cases were detected on magnetic resonance imaging only.

EA/TEF. Of these 13 cases, 10 were subsequently confirmed to have EA/TEF by detection of a pouch, one case was falsely ruled out on a targeted scan and two patients declined further investigation.

Overall, 3/69 cases scanned in the mid-trimester were diagnosed with an esophageal pouch on this scan, giving a detection rate of 4.3% for EA/TEF on the mid-trimester scan (Table 2). A further case with an esophageal pouch was detected only on MRI during the mid-trimester, giving a 5.8% detection rate for EA/TEF during the mid-trimester when considering both modalities.

Contribution of third-trimester scan to EA/TEF suspicion and detection

During the third trimester, 18/41 (43.9%) cases scanned that had not previously been suspected for EA/TEF were considered suspicious, owing to either a small/absent stomach or worsening polyhydramnios. Of these 18 cases, 13 were confirmed to have an esophageal pouch and five were not confirmed (two were assumed to have EA/TEF without pouch detection, two were falsely ruled out and one patient declined further investigation) (Figure 2).

In total, 84.0% (21/25) of prenatally detected cases were diagnosed during the third trimester, of which 19 were observed on ultrasound and two were detected only on MRI, giving a 33.9% (19/56) esophageal pouch detection rate for the third-trimester scan and a 37.5% (21/56) esophageal pouch detection rate for both modalities combined during the third trimester (Table 2).

Rate and timing of detection of EA/TEF and of sonographic signs

Table 3 shows the rate and timing of detection of each of the three sonographic signs of EA/TEF in the whole series. Polyhydramnios was the most common sign of EA/TEF and appeared in two-thirds of the group, as early as 21 weeks. A small/absent stomach was apparent in just over a third of the series, usually by the mid-trimester, but in some cases as early as 15.1 weeks. An esophageal pouch, the only direct diagnostic sign of EA/TEF, was present in about 33% of cases and was usually detected during the third trimester and, in a minority of cases, as early as 22.6 weeks. Approximately 30% of cases did not have any sonographic signs throughout pregnancy.

Multivariate logistic regression analysis of sonographic signs

We performed multivariate stepwise logistic regression analysis to assess the association between the indirect sonographic signs of EA/TEF (polyhydramnios, severe polyhydramnios and small/absent stomach) and the prenatal detection of EA/TEF. Severe polyhydramnios was removed during stepwise regression due to collinearity. A small/absent stomach and polyhydramnios were both strongly associated with the prenatal detection of EA/TEF, with odds ratios (ORs) of 10.4 (95% CI, 2.99–36.45; $P < 0.0001$) and 10.7 (95% CI, 1.22–95.3; $P = 0.03$), respectively.

Performance of MRI in pouch detection

Seventy-six percent (19/25) of the prenatally diagnosed cases were first detected on ultrasound, and the remaining 24.0% (6/25) on MRI. After the primary detection of an esophageal pouch on MRI, a pouch was also

Table 3 Detection and timing of detection of sonographic signs of esophageal atresia and/or tracheoesophageal fistula in 75 affected fetuses

Sonographic presentation	*Value*
Polyhydramnios	50 (66.7)
GA at detection (weeks)	30.4 (21.0–39.1)
Small/absent stomach	27 (36.0)
GA at detection (weeks)	23.6 (15.1–37.4)
Esophageal pouch	
Detected on US	22 (29.3)
Detected on MRI	12/18 (66.7)
Detected on US or MRI	25 (33.3)
GA at detection (weeks)	31.5 (22.6–38)
Primary detection modality	
US	19/25 (76.0)
MRI	6/25 (24.0)
Sonographic signs absent	23/75 (30.7)

Data are given as *n* (%), median (range) or *n*/*N* (%). GA, gestational age; MRI, magnetic resonance imaging; US, ultrasound.

Table 2 Suspicion and diagnosis of esophageal atresia and/or tracheoesophageal fistula (EA/TEF) on each anomaly scan in 75 affected fetuses

Sonographic presentation	*Early anomaly scan (n = 55)*	*Routine mid-trimester scan (n = 69)*‡	*Third-trimester scan (n = 56)*‡	P
Polyhydramnios	0 (0)	11 (15.9)	39/45 (86.7)	< 0.0001
Small/absent stomach	2 (3.6)	13/67 (19.4)	12/41 (29.3)	0.003
EA/TEF suspected*	2 (3.6)	13/67 (19.4)	18/41 (43.9)	< 0.0001
EA/TEF detected†	0 (0)	3 (4.3)§	19 (33.9)¶	< 0.0001

Data are given as *n* (%) or *n*/*N* (%). *EA/TEF was considered suspected if a small/absent stomach and/or worsening polyhydramnios was identified on ultrasound. †EA/TEF was considered detected if an esophageal pouch was identified on ultrasound or magnetic resonance imaging (MRI); cases detected only by MRI are not included in the table. ‡Once a case was suspected, it was omitted from analysis of suspected EA/TEF on subsequent scans. §An esophageal pouch was detected only on MRI in a further case on the routine mid-trimester scan, giving a 5.8% EA/TEF detection rate at this scan when considering both modalities. ¶An esophageal pouch was detected only on MRI in a further two cases on the third-trimester scan, giving a 37.5% EA/TEF detection rate at this scan when considering both modalities.

demonstrated on ultrasound in 3/6 cases. All of the detected cases were first suspected on ultrasound, owing to either worsening polyhydramnios, a small/absent stomach or both. Overall, 18 pregnancies underwent MRI for suspected EA/TEF, of which 12 were diagnosed accurately, giving a 66.7% detection rate and a 33.3% false-negative rate for MRI.

Effect of scan setting on sonographic sign detection

The anomaly scans were performed either at a tertiary referral center or in a local clinic. In order to determine whether the different settings of the exams affected the rate of sonographic sign detection, we compared the outcomes of routine and focused scans for conditions other than EA/TEF between the settings (Table 4). There was no statistically significant difference in the rate of absent signs for EA/TEF between the groups. The rate of polyhydramnios was also similar. Despite this, there were higher detection rates of a small/absent stomach and an esophageal pouch in cases assessed at the referral center than in those assessed at a local clinic. The diagnosis of EA/TEF by the detection of an esophageal pouch on ultrasound was established in 29% of cases assessed at the tertiary center compared with none of those assessed at a local clinic ($P = 0.01$).

Effect of type of EA/TEF on sonographic sign detection

Differences in the prenatal manifestation between Type-A cases (pure EA, without a fistula connecting the esophagus and the trachea) and cases involving a fistula (Types B–E) were assessed (Table 5). As expected, Type A was diagnosed prenatally at a significantly higher rate than were Types B–E. All six Type-A cases exhibited all three sonographic signs. On the other hand, in more than a third of cases involving a fistula, sonographic signs were absent. Only two cases were Type-E fistulas, the majority of cases being Type C.

Effect of associated anomalies on prenatal diagnosis of EA/TEF

Table 6 presents accompanying anatomic/genetic abnormalities and perinatal outcomes of the whole series.

Table 4 Detection of sonographic signs of esophageal atresia and/or tracheoesophageal fistula (EA/TEF) in 44 affected fetuses, according to whether the scans were performed at a local clinic or at a referral center

Sonographic sign	*Referral center scan* (n = *24*)	*Local clinic scan* (n = *20*)	P
Polyhydramnios	13 (54.2)	10 (50.0)	0.78
Small/absent stomach	9 (37.5)	2 (10.0)	0.036
Esophageal pouch	7 (29.2)	0 (0)	0.01
No sonographic signs	9 (37.5)	10 (50.0)	0.4

Data are given as *n* (%). Only routine scans and focused scans for conditions other than EA/TEF, such as other anomalies, growth monitoring and Doppler investigation are included.

Overall, 41 patients underwent genetic testing (seven microarray only, 19 karyotyping only and 15 both karyotyping and microarray). Of these, 7.3% of cases were diagnosed with a clinically significant genetic abnormality (two cases of trisomy 18 and one case of 22q11.23 duplication). EA/TEF was isolated in about 40% of cases and over half were VACTERL (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal abnormalities and limb abnormalities) associated. Around 8% of cases had non-VACTERL associated anomalies. There was no significant difference in the prenatal diagnosis rate of EA/TEF between cases with an associated anomaly and isolated cases (28.3% *vs* 41.4%; $P = 0.316$).

Our series of 76 cases showed a male predominance for EA/TEF, with a 1:1.6 ratio of females to males. Over 45% (33/71) of liveborn cases were born preterm and 40% had fetal growth restriction. Twenty-two percent (17/76) of cases were a cotwin (eight from monochorionic and nine from dichorionic pregnancies discordant for EA/TEF).

Table 5 Detection of sonographic signs of esophageal atresia and/or tracheoesophageal fistula in 70 affected fetuses, according to type of defect (Type A (pure atresia) or Types B–E (involving a fistula))

Sonographic sign	*Type A* (n = *6*)	*Types B–E* (n = *64*)	P
Polyhydramnios	6 (100.0)	39 (60.9)	0.082
Small/absent stomach	6 (100.0)	17 (26.6)	0.001
Esophageal pouch	6 (100.0)	14 (21.9)	< 0.0001
No sonographic signs	0 (0)	23 (35.9)	0.08

Data are given as *n* (%).

Table 6 Associated genetic and anatomical abnormalities and perinatal outcome in 76 fetuses with esophageal atresia and/or tracheoesophageal fistula (EA/TEF)

Variable	*Value*
Aneuploidy	2/41 (4.9)‡
Clinically significant CNV	1/21 (4.8)§
Isolated EA/TEF	30 (39.5)
VACTERL associated	40 (52.6)
Non-VACTERL anomalies	6 (7.9)
EFW < 10th centile	30 (39.5)
GA at delivery (weeks)*	36.6 (34.4–38.6)
Preterm birth	33/71 (46.5)
BW (g)*	2344 (1820–2865)
BW centile*	16 (4–34)
BW < 10th centile*	29/71 (40.8)
Male gender	47 (61.8)
Termination of pregnancy	3 (3.9)
Fetal demise	2 (2.6)
Neonatal death*†	6/71 (8.5)
Age at EA repair (days)*	3 (2–6)

Data are given as *n*/*N* (%), *n* (%) or median (interquartile range). *In liveborn cases ($n = 71$). †Two neonatal deaths were due to trisomy 18, one was due to extremely low birth weight (BW), one was due to other severe anomalies and two were due to operational and TEF-related complications. ‡Both cases had trisomy 18. §Case of 22q11.23 duplication. CNV, copy-number variant; EFW, ultrasound estimated fetal weight; GA, gestational age; VACTERL, vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal abnormalities and limb abnormalities.

DISCUSSION

This is the first study to address the timing of appearance of a small/absent stomach in fetuses with EA/TEF. Previous studies were performed in populations of fetuses with EA/TEF that were first scanned in the mid-trimester[7,15], and gestational age at the appearance of a small/absent stomach was not noted[5,15,16]. As anomaly scans do not include assessment of the esophagus and as polyhydramnios develops only after 20 weeks' gestation, the only feature discernible at this stage is a small/absent stomach.

In our study, the early anomaly scan raised suspicion for EA/TEF in only 3.6% (2/55) of cases. One of these cases was diagnosed with an esophageal pouch at 22.6 weeks, which is the earliest documented in the literature[5,12,17].

Whether a small/absent stomach was truly not present or merely perceived as such is difficult to determine retrospectively. Anomaly scans do not incorporate gastric-bubble measurements[14].

Theoretically, it is feasible to detect or rule out most suspected cases of EA/TEF on the mid-trimester scan because, by this stage, fetal swallowing with fluid propagation through the esophagus has been reported to be demonstrable in over 90% of cases[11]. In our series, the mid-trimester and third-trimester scans performed better at suspecting and detecting EA/TEF than did the early anomaly scan ($P < 0.00001$). On the routine mid-trimester scan, the suspicion rate (small/absent stomach and/or worsening polyhydramnios) was 19.4% and the detection rate (esophageal pouch) was 4.3%. On the third-trimester anomaly scan, 43.9% of cases were suspected (either small/absent stomach or worsening polyhydramnios) and 33.9% were detected.

The setting of the anomaly scan (local clinic or tertiary referral center) played an important role. Even when excluding suspicious cases referred for a targeted scan, cases scanned at a tertiary center were diagnosed significantly more often. Possible explanations for this phenomenon could be differences between the settings in the level of expertise, the type of ultrasound machine used and the length of the exams. Our observation is in agreement with the findings of a previous study comparing the detection rate of EA/TEF between district hospitals and referral centers[9]. Therefore, it is imperative that suspicious cases are referred to an appropriate tertiary center for ongoing prenatal investigation and care.

The presence of a TEF lowered the chance of prenatal diagnosis of EA/TEF and detection of its indirect signs (Table 5). This is most probably due to an ameliorating effect of the fistula on polyhydramnios and small/absent stomach, both of which were found to have an OR of around 10 for the prenatal diagnosis of EA/TEF. This phenomenon has also been observed in previous studies[7,15,18,19].

We conclude from our data that an early anomaly scan in its current form can suspect only a minority of cases of EA/TEF. A larger proportion of cases are suspected and diagnosed on the routine mid-trimester scan, but the vast majority of cases are detected on the third-trimester anomaly scan ($P < 0.00001$). As this is the first study to address the performance of the early anomaly scan in diagnosing EA/TEF, we have no previous data with which to compare our results. Our results show an overall prenatal detection rate (on ultrasound plus MRI) of 33.3% and a suspicion rate of 44.0%, which are similar to those reported in the published literature[2,5,20]. These rates are disappointingly low for such a significant anomaly. Most previous reports do not make a distinction between suspected cases (small/absent stomach or worsening polyhydramnios) and diagnosed cases (esophageal pouch). In a large European multicenter registry study including a total of 1222 cases of AE/TEF, the rate of prenatal detection ranged from 6.3% to 58.8% at different centers[2]. The study did not clarify which criteria were used for prenatal diagnosis and whether an esophageal pouch was mandatory for a definitive diagnosis. Only three previous studies referred to the demonstration of a pouch[12,16,19], one of which reported a sensitivity of 80% for prenatal ultrasound[12]. Contrary to our study, the largest of these studies considered polyhydramnios and/or small/absent stomach as diagnostic, despite the fact that only 0.002% of cases with polyhydramnios[21], 3–10.5% of cases with an absent stomach[22–24] and 33.3–37.5% of cases of both polyhydramnios and an absent stomach[23,24] have been reported to be due to EA/TEF.

Our series included eight cases that were suspected but not eventually diagnosed. Hence, there is room to improve our ability to diagnose an esophageal pouch. The complexity of esophageal-pouch identification arises from various causes. Firstly, visualization of the normally collapsed esophagus requires expertise and knowledge of the fetal neck and chest anatomy in order to be able to follow the course of the esophagus from the pharynx to the stomach. The scanner should be familiar with neighboring hypoechoic neck structures, such as the trachea and blood vessels, which may cause confusion when trying to identify the esophagus. Secondly, the detection of a pouch or a patent esophagus takes time and effort[12,17,25] and requires a supine fetal position with a slight extension of the neck. As the prevalence of this malformation is low, scanning for the propagation of fluid in the esophagus should be performed selectively in higher-risk patients, such as those with polyhydramnios and a small/absent stomach.

As shown in our study, just over 30% (23/75) of cases with EA/TEF did not display any sonographic signs and therefore could not have been suspected prenatally. Of these 23 non-suspicious cases, 12 had accompanying VACTERL anomalies. In fact, 52.6% of cases in our series were VACTERL associated. Therefore, if we were to scan actively for an esophageal pouch in cases presenting with VACTERL-associated anomalies, we could potentially improve the detection of EA/TEF in cases with absent sonographic features. In order to further improve screening for this major anomaly, we could consider adding the esophagus to routine scans. Feasibility studies of esophageal anatomy and functional assessment as early as the first trimester have shown this

to be achievable using high-resolution linear/curvilinear transducers[11,26]. Currently, the esophagus is not included in international guidelines for the anomaly scan[14].

Conclusions

Prenatal detection of EA/TEF is not feasible before the late second trimester. The early and routine mid-trimester anomaly scans in their current form perform poorly as a screening or diagnostic test for EA/TEF. The detection rates of all three sonographic signs of EA/TEF increase with advancing pregnancy, peaking in the third trimester. In the future, we could consider routine assessment of the fetal esophagus during anomaly scans in order to better screen for this major anomaly.

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