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Accuracy of prenatal detection of tracheoesophageal fistula and oesophageal atresia☆

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ABSTRACT

Aims: This study aims to determine the rate of prenatal detection of tracheoesophageal fistula and oesophageal atresia (TOF/OA), by identifying a small or absent stomach bubble with or without polyhydramnios, on the prenatal ultrasound scans (USS).
Methods: A retrospective study of prenatal ultrasound findings of babies with a prenatal and postnatal diagnosis of TOF/OA born between 1st January 2004 and 31st December 2013 was undertaken.
Results: A total of 58 babies were born with TOF/OA. 40% of mothers had their prenatal investigations performed within our tertiary centre, and the remaining 60% had their antenatal care at their local district general hospital (DGH). The overall sensitivity for prenatal USS was 26%, with a specificity of 99% and a positive predictive value (PPV) of 35%. However, the sensitivity of the prenatal USS within the tertiary centre was significantly higher at 57%, while only 2 cases were detected prenatally in the DGHs. Polyhydramnios was seen in 67% of mothers that had a prenatal diagnosis of TOF/OA and its presence did significantly increase the positive predictive value of prenatal USS (from 35% to 63%). Of those that were postnatally diagnosed, 21% had prenatal polyhydramnios. There was no significant difference in postnatal outcomes between those that were prenatally diagnosed and those that were postnatally diagnosed.
Conclusion: Prenatal diagnosis of TOF/OA remains challenging. However within a specialist centre the accuracy of successful prenatal detection can be significantly improved. This is beneficial both for prenatal counselling of families and for planning appropriate perinatal and postnatal care for the baby.

Tracheoesophageal fistula and oesophageal atresia (TOF/OA) is a rare congenital abnormality with an incidence estimated at approximately 1 per 3500–4000 live births [1]. The condition can be detected prenatally during the routine fetal anomaly ultrasound scan (USS) usually performed at 20 weeks gestation. Features of a small or absent gastric bubble especially in association with polyhydramnios should raise suspicion of TOF/OA (Fig. 1). In 50% of cases TOF/OA can be associated with other congenital anomalies or syndromes, such as VACTERL association, CHARGE association and chromosomal anomalies e.g. Trisomy 18 [2]. Therefore, a prenatal suspicion of TOF/OA should also alert the fetal medicine team to search for features of other associated anomalies. Unfortunately the ultrasound features for TOF/OA are non-specific, subjective and sometimes transient in nature [3]. For these reasons the accuracy of prenatal detection has previously been described to be poor with a high rate of false positive results [1,3–5].

Effective prenatal recognition of TOF/OA and other congenital anomalies allows time to plan appropriate and prompt postnatal management, hence avoiding risks associated with a delayed diagnosis. It can then be extrapolated that a prenatal diagnosis has the potential to improve postnatal clinical outcomes [6]. Additionally it benefits families by allowing for effective prenatal counselling [7]. Conversely a false positive result can lead to unnecessary anxiety and stress for families.

This study aims to determine the current accuracy of prenatal detection, both in general prenatal care and within a specialist tertiary centre. Over the last decade there has been significant progress in many aspects of fetal medicine, including technology, expertise and availability. By comparing this data to a previously published study from the same unit a decade ago, we aim to determine whether accuracy of prenatal detection of TOF/OA has improved. In addition, comparison of those babies with a prenatal diagnosis and those with a postnatal diagnosis aimed to identify whether there are any significant differences in these two patient groups in order to better guide future prenatal counselling and postnatal management.

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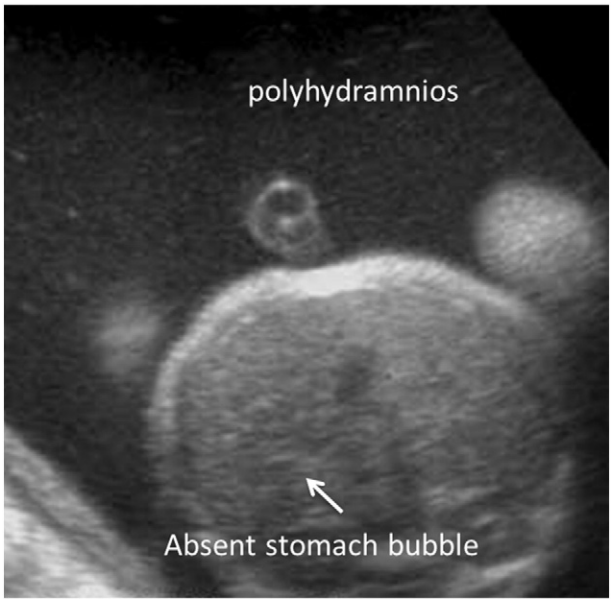

Fig. 1. Image from a prenatal ultrasound demonstrating an absent stomach bubble and polyhydramnios, findings consistent with a diagnosis of a TOF/OA.

1. Methods

A retrospective study of all babies with either a prenatal suspicion of TOF/OA or a postnatal diagnosis of TOF/OA that were born between 1st January 2004 and 31st December 2013 was undertaken.

Cases with a prenatal suspicion were identified from the local congenital anomaly register and included all mothers within the local catchment area and those cases referred for second opinion scans from the district general hospitals (DGHs). Fetal anomaly scans were performed between 20 and 24 weeks gestation. A prenatal diagnosis of TOF/OA was based on two prenatal scans performed on different dates reporting a finding of a small or absent stomach bubble, with or without associated polyhydramnios. All mothers with a prenatal suspicion of TOF/OA were referred to and seen in the combined surgical fetal medicine clinic at the regional tertiary centre and were included in this study. The location of prenatal care was determined by the location of the first anomaly scan showing suspicion of TOF/OA and was documented as either within the tertiary centre or the DGH.

In addition all neonates diagnosed with a TOF/OA and treated at our tertiary centre during the same time period were identified using the neonatal surgical database and clinical coding systems. TOF/OA was diagnosed by failure to pass a nasogastric tube into the stomach. Patients with a diagnosis of H-type tracheoesophageal fistula were excluded. Clinical details, including prenatal scan results, epidemiology and postnatal outcomes were obtained from the clinical notes. Cases were cross referenced and duplicates were excluded. All babies were treated in the paediatric surgery unit, by one of six consultants performing these operations.

The two groups of patients were then correlated and analysed to determine the true positives, true negatives, false positives and false negatives. A contingency table was then created to calculate the sensitivity, specificity, positive predictive valve (PPV) and negative predictive value (NPV) for prenatal USS at detecting TOF/OA.

The two patient groups (prenatally detected TOF/OA and postnatally detected TOF/OA) were then compared to determine any difference in patient characteristics, clinical features and outcomes. Statistical analysis was performed with a Z test for proportional data and a Mann–Witney U test for continuous data. Statistical significance was defined as a p-value of <0.05.

2. Results

Within the tertiary centre the number of pregnancies undergoing prenatal care is approximately 7050 per year, while within the DGHs situated in the referring region it is approximately 41,000 per year. Therefore the total number of pregnancies undergoing prenatal care within the tertiary centre during this 10 year study period was approximately 70,500. Of all of these prenatal scans, both in the tertiary centre and DGHs, approximately 10% demonstrate polyhydramnios.

Review of the local fetal medicine congenital anomaly register for this study period identified 43 fetuses in which a prenatal diagnosis of TOF/OA was suspected. Of these babies, 15 were confirmed to have TOF/OA at birth. Review of the neonatal surgical database identified 58 babies diagnosed with TOF/OA. These cases were correlated with the fetal medicine register, which confirmed that 15 babies had a prenatal diagnosis of TOF/OA and 43 babies were diagnosed with TOF/OA postnatally with no suspicion on prenatal USS. Therefore prenatal USS resulted in 15 true positives with 28 false positives and 43 false negatives. As the condition is rare the true negative rate, and also therefore specificity, are difficult to determine because of the large number of true negative scans. This has been approximated at 70, 442; the total number of prenatal scans being performed in the tertiary centre during the study period excluding those babies either with a prenatal suspicion of TOF/OA or a subsequent diagnosis of TOF/OA. The overall sensitivity of prenatal USS in detection of TOF/OA was calculated at 26% with a specificity of 99% and a PPV of 35% (Table 1).

3. Location of care

In total there were 58 babies with a confirmed diagnosis of TOF/OA. Thirty-five (60%) of these babies had their prenatal care within the district general hospitals (DGHs). However, only 2 of these mothers had a successful prenatal diagnosis of TOF/OA. The remaining 23 (40%) babies had their prenatal care within the tertiary centre and 13 of these were detected prenatally (Fig. 2). The sensitivity of prenatal USS was therefore significantly higher for those scans performed in the tertiary centre compared to the DGHs; 57% and 6% respectively ($p < 0.01$).

4. Polyhydramnios

Polyhydramnios was seen in 16/43 (37%) of mothers that had a prenatal suspicion of TOF/OA. Of these babies, that had USS findings of small or absent stomach and polyhydramnios, 10 babies were subsequently confirmed to have a TOF/OA postnatally, while there were 6 false positive results. Therefore the PPV for these findings in combination was 63% and was significantly higher than a small/absent stomach without polyhydramnios ($p = 0.027$). Of those that were postnatally diagnosed, 9/43 (21%) had prenatal polyhydramnios with what was thought to be a normal stomach and therefore they were not identified as having a prenatal suspicion of TOF/OA. In total, polyhydramnios was present in 9/58 (33%) of all pregnancies where a TOF/OA was confirmed.

5. Associated anomalies

TOF/OA was an isolated finding in 33/58 (67%) of babies. Associated anomalies were present in the remaining 25 babies, with the most common anomalies being VACTERL association (10), cardiac abnormalities (5), CHARGE association (2), Trisomy 18 (2), and anorectal malformations (2). Of those babies with a postnatal diagnosis of TOF/OA, 16 (37%) had associated anomalies. Three babies had associated anomalies identified on their

Table 1
Contingency table demonstrating the sensitivity, specificity, positive predictive value (PPV) and negative predictive value (NPV) for prenatal scans in detecting TOF/OA.

	TOF/OA	No TOF/OA	
Prenatal scan suspicious	15 (13)	28	PPV 35%
Normal prenatal scan	43 (10)	70 448	NPV 99%
	Sensitivity 26% (57%)	Specificity 99%	

Brackets indicate the numbers for those mothers undergoing their prenatal care within the tertiary centre rather than the DGH.