

First trimester findings of combined duodenal and oesophageal atresia with a trachea-oesophageal fistula

Tegan Sullivan¹

BBiomedSc(Newcastle),
GradDipSon(RMIT), AMS

Meiri Robertson^{1,2}

MBChB
BScMedSc(Hons)Human
genetics

Lavinia Hallam³

BSc (Hons) (Wales) MB
BCh (Wales) FRCPath (UK)
FRCPA (Aus) Dip Foren
Path (Aus) Cert Health
Serv Man (WA)

¹Fetal Medicine Unit
Women and Children's
Health Services
The Canberra Hospital
Department of Obstetrics
and Gynaecology
Garran
Australian Capital
Territory
Australia

²Australian National
University Medical School
Canberra
Australian Capital
Territory
Australia

³ACT Pathology
The Canberra Hospital
Garran
Australian Capital
Territory
Australia

Correspondence to email
tegan.sullivan@act.gov.au

Abstract

This case demonstrates the ability to diagnose a significant duodenal atresia coexisting with an oesophageal atresia with associated tracheo-oesophageal fistula within the first trimester, however the mechanisms underlying the sonographic features remain uncertain.

Keywords: duodenal atresia, oesophageal atresia, tracheo-oesophageal fistula.



Figure 1: Dilated fetal stomach at 12 weeks.

Duodenal atresia (DA), is a developmental anomaly caused by failure of duodenal recanalisation at eight weeks gestation, and has an estimated incidence anywhere from 1:10,000 to 1:40,000 live births. There is a strong association of isolated DA and Trisomy 21, with a 30% incidence of an affected fetus¹. Prenatal sonographic findings of DA have been well established; the typical “double bubble” seen on ultrasound is due to gastrointestinal obstruction – the combination of a distended stomach and distension of the duodenum proximal to the atresia. This is typically found within the second, and more commonly third trimester of pregnancy.

Isolated oesophageal atresia (OA) on the other hand, is a challenging diagnosis to make prenatally, affecting between one in 2500 to 4000 newborns². This is considered to arise from abnormal foregut development between 4–8 weeks gestation. There

is a high association between OA and additional abnormalities (30–50% of cases)², most commonly seen as part of the VACTERL spectrum (vertebral, anorectal, cardiac, tracheo-oesophageal, renal and limb defects). OA is also associated with abnormal karyotype; while Trisomy 21 is an uncommon association, it has been reported⁴. Sonographic findings include the combination of small or absent fetal stomach, polyhydramnios and “upper pouch sign” of the proximal oesophagus.

There is a well-described association between duodenal atresia and oesophageal atresia (with a tracheo-oesophageal fistula (TOF)), with 6% of OA having an associated DA². This diagnosis is associated with significant mortality and morbidity⁵. Isolated oesophageal atresia *without* a TOF is rare (seen in only 8% of all OA)⁶, however there are multiple case reports of combined



Figure 2: Double bubble at 15 weeks.

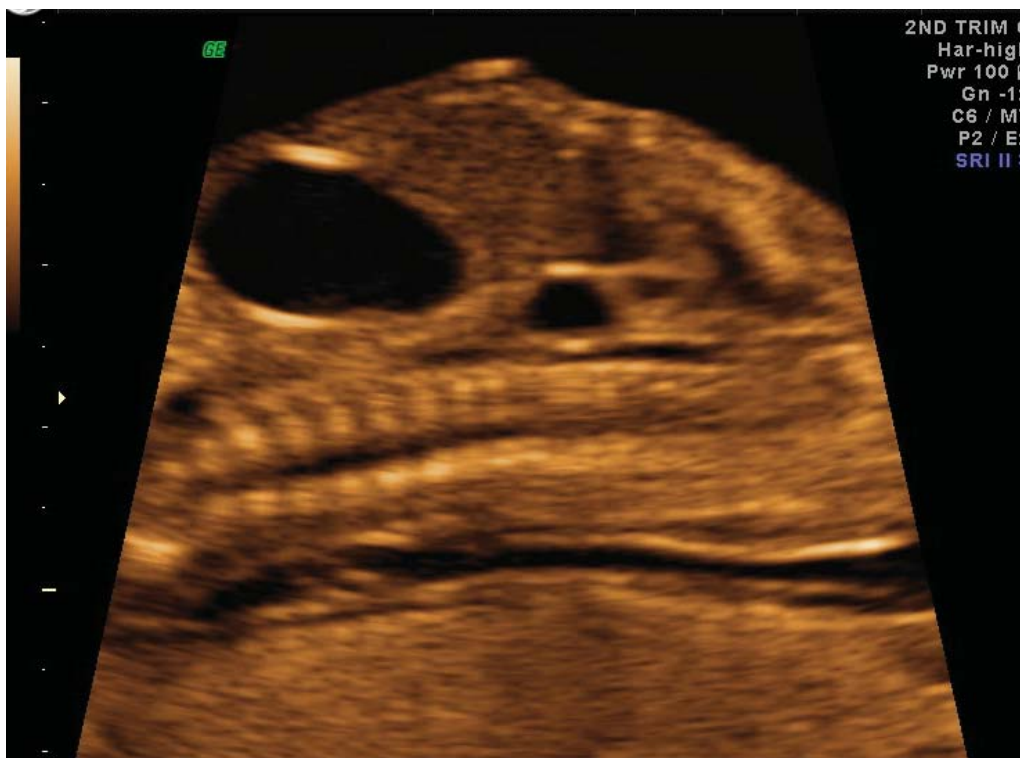


Figure 3: Sagittal section demonstrating dilated stomach and oesophagus.

DA and OA without a TOF. Mitani, *et al.*³ identified 11 cases (including their own) of prenatally diagnosed concomitant DA and OA without TOF within the English literature. Gestational age at diagnosis ranged from 12–36 weeks, which included two cases of first trimester detection^{1,7}. To the best of our knowledge, this case of combined duodenal and oesophageal atresia *with* an associated trachea-oesophageal fistula diagnosed in the first trimester, is the first case reported in the English literature.

A 32-year-old Gravida 4, Para 1 presented to our tertiary referral unit at 12 weeks for routine nuchal translucency screening. Nuchal translucency measured 1.69 mm, with the combined

risk of Trisomy 21 of 1:168 (abnormal biochemistry – β HCG 2.60 MoM, PAPP-A 0.61). The patient declined invasive testing. First trimester fetal anatomy revealed a significantly dilated fetal stomach (Figure 1), along with a dilated cystic structure noted posteriorly in the upper chest. Follow-up imaging at 15 weeks confirmed a double bubble with the additional finding of a cystic structure in the upper chest displacing the descending aorta anteriorly (Figures 2–4). The presence of duodenal atresia and probable coexisting oesophageal atresia was suspected. An uncomplicated amniocentesis was performed with fast Fluorescence *In situ* Hybridisation (FISH) revealing Trisomy 21.

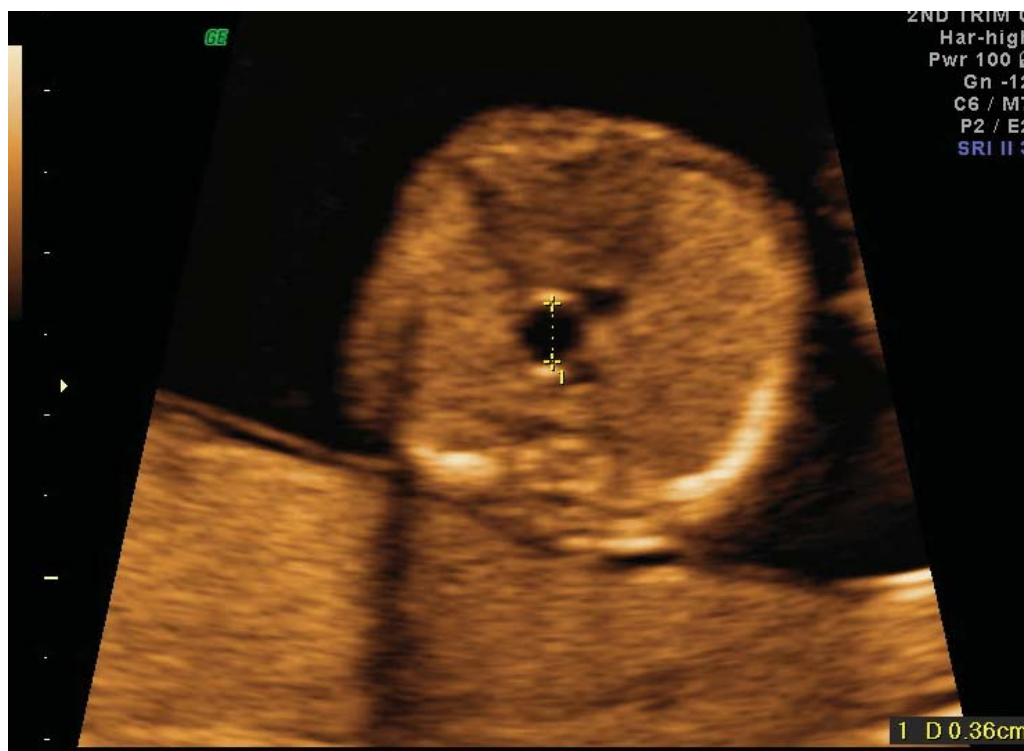


Figure 4: Dilated oesophagus in the upper chest displacing the descending aorta anteriorly.

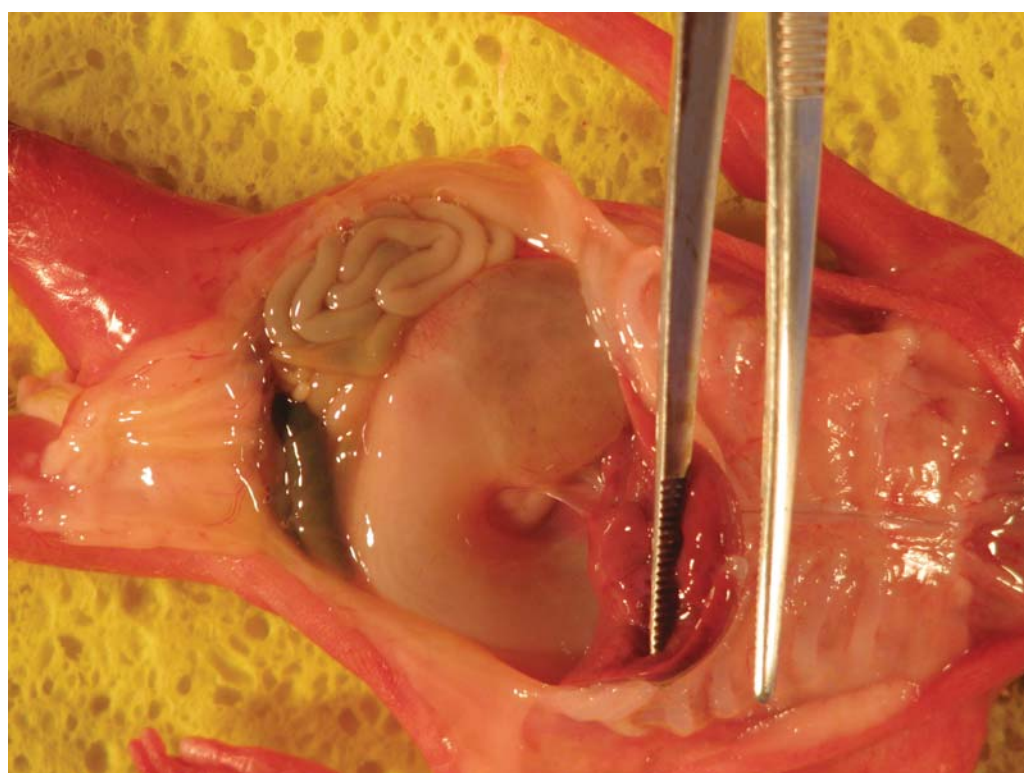


Figure 5: Abdominal organs showing enlarged stomach and first part of the duodenum with relatively empty distal bowel. There is also splaying of the inferior costochondral margins.

The pregnancy was interrupted at 18 weeks gestation and autopsy confirmed the ultrasound findings (Figure 5). The proximal oesophagus ended blindly (Figure 6); the distal end communicating into the right proximal bronchus (distal trachea-oesophageal fistula). Autopsy images show a markedly distended stomach and duodenum. There was the additional finding of an aberrant right subclavian artery.

This case is unusual in many ways. First, the association of

a combined DA and OA in a Trisomy 21 fetus is an uncommon finding⁴. Second, the sonographic features seen in this case are normally associated with a DA/EA complex without a TOF. Mitani, *et al.*³ theorise that OA without TOF is prenatally diagnosed with a small or absent stomach, but when combined with DA, the stomach and duodenum become markedly distended with secreted gastric and duodenal juices. Tsukerman, *et al.*⁸ believe that this combination leads to a closed loop of bowel

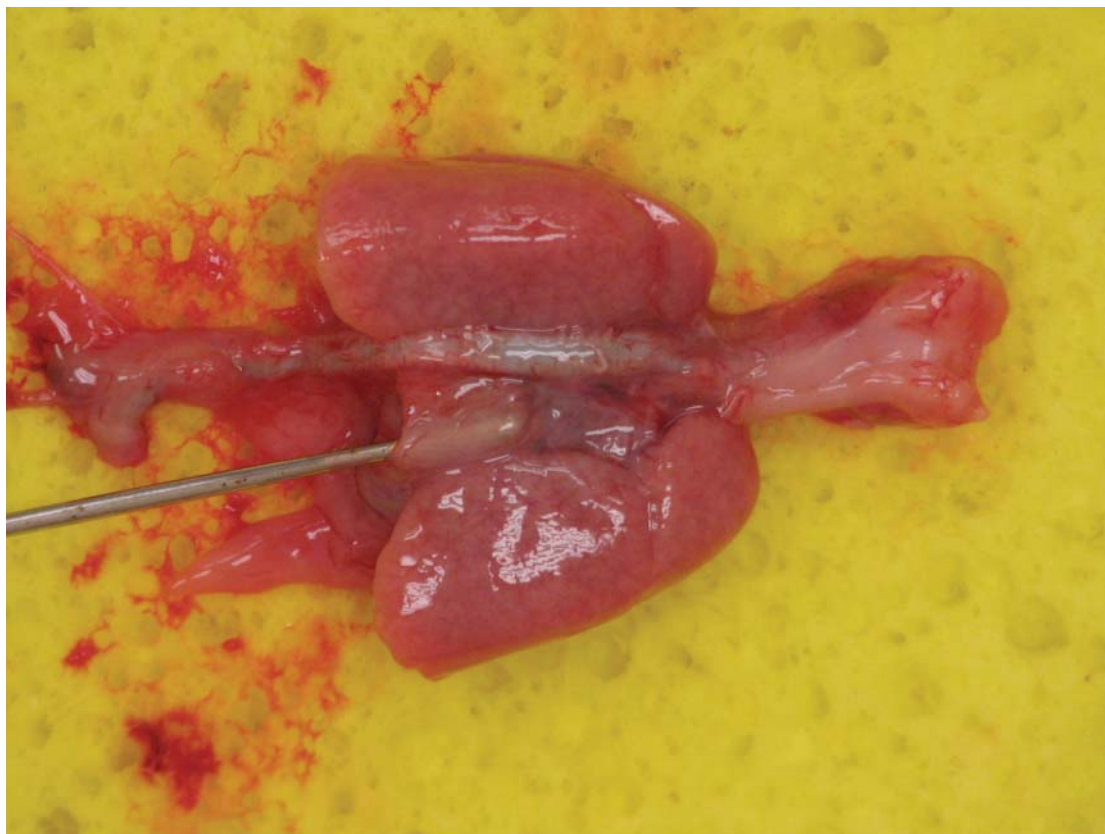


Figure 6: Demonstration of tracheo-oesophageal fistula with probe in the lower oesophagus.

that involves the distal oesophagus, stomach and duodenum.

Last, the findings of DE/OA with TOF are normally seen at a much later gestation, due to the mechanics of fetal swallowing and gastrointestinal secretions. Nicolaides, *et al.*⁹ report that sonographically demonstrable (and therefore significant) oesophageal distension is a rare finding in the second and third trimesters, and the incidence in the first trimester is unknown. It is believed that fetal swallowing in the first trimester would not contribute to stomach and oesophageal dilatation and the stomach is therefore distended by gastrointestinal and pancreatic secretions alone. However, the secretions produced by a first trimester fetus are unlikely to be sufficient to cause significant dilation at this early gestation. The findings in our case lead us to propose another mechanism; the trachea-oesophageal fistula was constricted, therefore providing mechanical obstruction. This produced ultrasound features of a true obstruction both at the oesophageal “inlet” and duodenal “outlet” without a TOF.

This case demonstrates the ability to diagnose a significant duodenal atresia coexisting with an oesophageal atresia with associated tracheo-oesophageal fistula within the first trimester, however the mechanisms underlying the sonographic features remain uncertain.

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