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# Increased fetal nuchal translucency: possible association with esophageal atresia

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## ABSTRACT

*We describe a case in which in-utero diagnosis of an esophageal atresia with a tracheo-esophageal fistula in the third trimester followed the finding of an increased nuchal translucency in the first trimester and suggest a mechanism by which these two findings might be associated.*

## INTRODUCTION

Nuchal translucency thickness (NT) measurement is a sensitive method of screening for fetal aneuploidy[1]. It has also been found that an increased nuchal translucency may be associated with a variety of fetal abnormalities in addition to fetal chromosomal disorders[2], including cardiac defects[3] a variety of genetic syndromes and other structural defects.

Here we describe a case in which in-utero diagnosis of an esophageal atresia with a tracheo-esophageal fistula (TOF) in the third trimester followed the finding of an increased nuchal translucency in the first trimester.

## CASE SUMMARY

A 32-year-old woman in her second pregnancy, the first having been uncomplicated, attended for nuchal translucency screening for fetal aneuploidy at 12 weeks of gestation. The NT measured 3.0 mm, resulting in an increased risk for fetal trisomy. The remainder of the anatomical survey was normal. After counseling the parents elected to undergo fetal karyotyping by chorion villus sampling. The fetal karyotype was normal. In view of the association between increased NT and cardiac defects in addition to a full anatomic survey at 20 weeks of gestation, fetal echocardiography was performed at 14 and 20 weeks. All these subsequent investigations were normal.

At routine antenatal assessment at 31 weeks of gestation the symphysis-fundal height was increased (40 cm). Ultrasonography confirmed polyhydramnios with a four-quadrant amniotic fluid index (AFI) measuring 33 cm. There were no obvious fetal defects. Glucose challenge test and maternal infection screen were negative.

By 32 weeks of gestation the AFI had increased to 38 cm. On this occasion the stomach remained only partially filled despite intermittent observation of the fetus over a prolonged period. This suggested that the etiology of the polyhydramnios might be an esophageal atresia with coexistent TOF. At 36 weeks of gestation the baby was delivered by Cesarean section for transverse lie and increasing polyhydramnios.

Esophageal atresia was confirmed at birth and at 24 h of age the baby underwent surgical repair of the defect. The operative findings were of a short atretic defect in the esophagus allowing primary anastamosis of the proximal and distal ends under only moderate tension with, in addition, a single TOF at the level of the carina which was divided.

## DISCUSSION

Among the mechanisms implicated in the etiology of transiently increased nuchal translucency are cardiac failure, anomalous lymphatic drainage and compression of the superior mediastinum. Such findings may be found in association with diaphragmatic hernias or asphyxiating thoracic dystrophies[2]. In this case esophageal atresia and TOF were associated with increased NT in the first trimester.

Polyhydramnios in cases of esophageal atresia results from a reduced turnover in amniotic fluid as a consequence of the esophageal obstruction. Such an effect exists whether there is a coexisting TOF or not. Collection of fluid in the upper esophagus, rather than the normal passage of amniotic fluid through it, may result in esophageal distension[4]. Significant, and therefore sonographically demonstrable, esophageal distension is not a common finding in the second and third trimesters and the

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incidence in the first trimester is unknown. In our hypothesis we speculate that this might occur.

At the opening of the esophagus its posterior wall is typically thinned and is the site of the pulsion pharyngesophageal or Zenker's diverticulum of adult life. In addition the esophagus of the neonate is relatively long compared with that of the adult, with the pharyngesophageal junction lying at the level of the third or fourth cervical vertebra rather than at the sixth[5]. It is therefore conceivable that esophageal distension would be most marked at its opening. In addition its more cephalad position anatomically could result in a degree of compression of the great vessels of the head and neck which lie to either side of the upper esophagus. By impairing venous return this might in turn result in an increased NT. With fetal growth and associated anatomic changes the impact of esophageal distension on the surrounding vessels would become less critical and the nuchal translucency resolve with increasing gestation.

Antenatal recognition of esophageal atresia or TOF may result in an improved outcome in such cases, by allowing the early neonatal initiation of appropriate measures such as the prevention of aspiration pneumonitis and allow early surgical repair. An increased NT in the first trimester is an important marker of a variety of fetal pathologic conditions. Such a finding requires systematic exclusion of known associated conditions. Even though it is known that other gastrointestinal atresias (e.g. duodenal atresia) are associated with an increased NT[2] further case studies will be required to establish whether esophageal atresia should be similarly categorized.

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