Antenatal Diagnosis of Esophageal Atresia with Tracheoesophageal Fistula

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The antenatal diagnosis of esophageal atresia by the presence of polyhydramnios and an absent stomach bubble is well documented. A significant number of esophageal atresias are associated with a tracheoesophageal fistula. In these cases, the diagnosis of esophageal atresia can be missed on a prenatal ultrasonogram since the fluid-filled stomach is seen. We present a case of esophageal atresia with a fistula diagnosed prenatally.

CASE REPORT

A 26 year old gravida 2, para 1 woman with a history of tracheal atresia detected at birth in the previous pregnancy was referred at 30 weeks of gestation for ultrasonography to rule out fetal anomalies. Ultrasonographic examination in the present pregnancy revealed polyhydramnios. Biparietal diameter and femur length were appropriate for gestational age. The fluid-filled stomach was seen (Fig. 1A) In view of polyhydramnios and the tracheal atresia in the previous pregnancy, the fetal neck and chest were imaged in detail. A dilated blind-ending proximal esophageal pouch was seen filling and emptying repeatedly (Figs. 1B and 2). Hence, a diagnosis of esophageal atresia with a tracheoesophageal fistula (type III, Ladd classification) was made. A male baby was delivered vaginally at term. A postnatal radiograph with a tube in the esophageal pouch confirmed the diagnosis of esophageal atresia and a tracheoesophageal fistula. During surgery, esophageal atresia with tracheoesophageal fistula was detected and corrected. The child had an uneventful postoperative period.

DISCUSSION

Esophageal atresia has a very poor prognosis postnatally, because aspiration of feeds and pharyngeal contents causes respiratory distress and bronchopneumonia, and leading to death. An antenatal diagnosis of esophageal atresia facilitates delivery in a hospital, where immediate postnatal surgery can be performed, and helps in the planning of immediate neonatal care. Precautions such as avoiding feeds to prevent aspiration help to reduce mortality.
Five major types of tracheoesophageal abnormalities have been described: (1) isolated esophageal atresia, (2) esophageal atresia with a fistula connecting the proximal portion of the esophagus with a trachea, (3) esophageal atresia with a fistula connecting the distal portion of the esophagus with the trachea, (4) esophageal atresia with a double fistula connecting both segments of the interrupted esophagus with the trachea, and (5) a tracheoesophageal fistula without esophageal atresia. The third variety is the most common and accounts for more than 90% of all cases.\(^1\)

Most cases of esophageal atresia and a tracheoesophageal fistula have been sporadic. There are a number of reports of more than one affected person in a family. However, since the actual prevalence of this defect in first-degree relatives is lower than the prevalence, genetic causes are not supported.\(^2\) In our case one sibling had tracheal atresia, the occurrence of which is well reported.\(^2\) Other birth defects are also reported in family members, but the most common is one of the VATER spectrum. A higher than expected frequency of birth defects was not found in other family members.\(^2\)

Polyhydramnios and a failure to visualize the stomach persistently on a prenatal scan suggest the diagnosis of esophageal atresia.\(^1,3-8\) These findings have been described in both isolated esophageal atresia and esophageal atresia with a tracheoesophageal fistula.\(^7,8\)

Based on these observations, the diagnosis is possible only in about 10% of the patients. In the remaining patients, the presence of gastric secretions or a tracheoesophageal fistula allows visualization of the stomach.\(^1\) A dilated blind-ending proximal esophagus showing alternate filling and emptying on a prenatal scan has been described in two cases of esophageal atresia.\(^7\) In both cases there was polyhydramnios and an absent stomach bubble. Our case had polyhydramnios, and the fluid-filled stomach was imaged. The diagnosis of esophageal atresia was made by visualizing the proximal dilated blind-ending esophageal pouch, showing alternate filling and emptying on real time. This observation helped us arrive at the correct diagnosis even though the stomach bubble was seen. Hence, it is a reliable and specific sign of esophageal atresia with or without a tracheoesophageal fistula.

A careful imaging of the fetal neck for evidence of a fluid-filled esophageal pouch should be done in all cases of polyhydramnios. This finding will help us diagnose esophageal atresia more accurately even in the presence of a tracheoesophageal fistula which is the most common type seen.
Figure 1 A, Transverse section of fetal abdomen shows the fluid-filled stomach. 
B, Coronal section of fetal neck and chest showing the blind-ending pouch of esophagus in the empty phase.
Coronal scan of neck and chest of fetus shows dilated blind-ending proximal esophageal pouch in the full phase.

REFERENCES


