Esophageal atresia + tracheo-esophagic fistula

Ultrasound findings

A GxxxPxxx pregnant woman presented at xxx weeks of gestation for a follow-up scan due to a persistent small/ stomach noted on previous evaluations. The stomach remained visible but small/ throughout the whole session (about 60 minutes). A careful evaluation of the neck on sagittal views showed a blind ending of the proximal part of the esophagus during fetal swallowing. Additionally, mild polyhydramnios was observed , with the deepest vertical pocket measuring xx cm.

Those which support the diagnosis

The rest of the fetal anatomy appeared unremarkable. Specifically, the fetal face, neck, and spine were normal. Normal fetal swallowing was observed. The diaphragm appeared intact +/- flattened everted, the kidneys were normal, and no cardiac defects were seen. The fetus was active, and the extremities showed normal tone with a good range of movements.

Pertinent negatives

There were no suggestive signs of fetal anemia or infections. The estimated fetal weight was on the XX centile, the maternal abdomen was soft and no contractions were noted.

The above findings strongly suggested esophageal atresia (EA) with tracheoesophageal fistula (TEF).

Is this an isolated finding?

We explained to the parents that the antenatal diagnosis of EA and TEF is challenging. However, the presence of a small or nonvisible gastric bubble, polyhydramnios, appearance of the diaphragm and the formation of an esophageal pouch significantly increases the risk . No associated anomalies were noted, which was reassuring.

Additional testing:

Blood testing: ie. genetic, infection, other maternal studies suggested

An amniocentesis for fetal cytogenetic studies (karyotyping and CGH-array) was offered and accepted. Additionally, the mother will be tested for gestational diabetes.

Ultrasound tests: ie. growth scans, need for echocardiogram if indicated.

An MRI test (if available) will be requested to better characterize the esophagus. Follow-up scans to monitor growth, well-being, and amniotic fluid will be scheduled. If polyhydramnios becomes severe, amniodrainage might be necessary, and there is a chance that preterm contractions and labor may occur.

Delivery issues, if any

Delivery is recommended at a tertiary referral hospital with a neonatal intensive care unit and pediatric surgery capability.

Post-natal evaluation

A neonatal alert will be raised. Diagnosis can be confirmed by placing a nasogastric tube at birth Additional imaging exams may be performed. If EA is confirmed, surgical treatment will be necessary. An individualized approach will be scheduled depending on the type of esophageal atresia/tracheoesophageal fistula (OA/TEF), its severity, the extent of the esophageal gap, and the expertise of the surgical team.

Prognosis

We have explained to the parents that chromosomal abnormalities (such as trisomy 21 and 18) are present in about 5-10% of cases of EA, and genetic syndromes such as VACTERL are also frequent. However, a low-risk first-trimester screening test and the absence of associated fetal anomalies are reassuring and decrease their likelihood.

Several risk classifications attempt to predict neonatal outcomes. Factors such as associated anomalies, pulmonary complications, and low birth weight can worsen the prognosis. The overall mortality ranges from 10 to 20%, mostly related to other structural abnormalities or chromosomal abnormalities. Genetic test results will be available in XX days.

The risk for fetal growth restriction and low birth weight is also increased. Polyhydramnios increases the risk of prematurity. Additionally, comorbidities after birth, such as postnatal growth retardation, recurrence of the tracheoesophageal fistula (TEF), gastroesophageal reflux, esophageal dysmotility, feeding difficulties, tracheomalacia, and recurrent respiratory infections, can occur. Therefore, long-term follow-up will be necessary. However, in most instances, patients treated for esophageal atresia (EA) have a good long-term functional course.

Additional consultations which are suggested

Counseling by a neonatologist and a pediatric surgeon will be arranged. Depending on the results of genetic testing and follow-up findings, other specialists might become involved.

Pre-pregnancy counseling for the next pregnancy

The etiology of esophageal atresia (EA) is multifactorial and remains unknown, although specific genetic defects and mutations have been described. Therefore, pre-pregnancy counseling by a geneticist is suggested for future events. Isolated, non-syndromic, chromosomally normal cases of EA carry a low recurrence risk of about 1%.