Congenital tracheoesophageal fistulas are a common congenital anomaly that is usually diagnosed prenatally or during the neonatal period. The condition is usually characterized by an esophageal atresia associated with a communication between the trachea and the esophagus. Maternal polyhydramnios is commonly seen in cases of esophageal atresia. The diagnosis can be confirmed prenatally by ultrasonography or postnatally by a chest x-ray or a computed tomography scan. Early surgical repair is recommended in all healthy infants.

Overview

A tracheoesophageal fistula, as the name implies, is a **communication between the trachea and esophagus**. Congenital tracheoesophageal fistulas are usually diagnosed at birth or soon thereafter.

Congenital tracheoesophageal fistulas can be classified into **five distinct types** based
on the site of the communication. The most common form, **esophageal atresia with distal tracheoesophageal fistula**, is seen in up to 86% of the cases.

The second most common form is an **isolated esophageal atresia** without a tracheoesophageal fistula. This type is seen in approximately 8% of the cases.

**Isolated tracheoesophageal fistulas** come in third place in frequency and is diagnosed in 4% of the cases.

The least common types of tracheoesophageal fistulas are **esophageal atresia with a proximal communication** and **esophageal atresia with proximal and distal communications**. These two types are responsible for 1% of the cases each.

**Epidemiology of Congenital Tracheoesophageal Fistulas**

The estimated incidence of congenital tracheoesophageal fistulas is **around 1 in 2,000 to 4,000 live births**. There are no racial differences and the incidence is equal in both genders.

Most newborns with congenital tracheoesophageal fistulas are diagnosed during the neonatal period because of the emergence of **severe complications**. In fact, the diagnosis of a congenital tracheoesophageal fistula is most commonly made at birth.

The prognosis of infants who had a congenital tracheoesophageal fistula surgically repaired is excellent with a 100%-survival rate. If early surgical repair is not possible, survival rate can drop by 10 to 20%.

Despite excellent survival rate, infants are at risk of developing several morbidities after the repair of the tracheoesophageal fistula. **Esophageal dysmotility**, **gastroesophageal reflux** and **dysphagia** are commonly seen.

**Recurrent pneumonia** is another common complication of tracheoesophageal fistulas.
**Tracheomalacia** has also been reported to be more common in infants who had a tracheoesophageal fistula.

**Etiology of Congenital Tracheoesophageal Fistulas**

*Certain genetic disorders* and *environmental exposures* have been linked to an increased risk of congenital tracheoesophageal fistulas. *Trisomies 18, 13 and 21* have been associated with tracheoesophageal fistulas.

Additionally, the use of the *decongestant imidazoline* in the first trimester has been linked to an increased risk of congenital tracheoesophageal fistulas. In most cases, however, a single etiology cannot be identified.

**Pathophysiology of Congenital Tracheoesophageal Fistulas**

Tracheoesophageal fistulas are usually associated with multiple anomalies that can affect the survival of the child. *Ventricular septal defects, patent ductus arteriosus, tetralogy of Fallot* and *atrial septal defects* are commonly seen in children with congenital tracheoesophageal fistulas.

*Genitourinary anomalies* such as renal agenesis, horseshoe kidney, polycystic kidney and ureteral or urethral malformations are also commonly associated with tracheoesophageal fistulas.

Tracheoesophageal fistulas can also be associated with a *duodenal atresia*, *malrotation of the gut, Meckel diverticulum* and *annular pancreas* as well as *vertebral defects, anal atresia* and *limb defects* (radial dysplasia, polydactyly, syndactyly). These associations suggest a common pathophysiology or etiology between these different malformations.

During *embryogenesis*, the *primitive foregut* gives rise to a *ventral diverticulum* which evolves into the trachea. This event usually starts happening in 4 to 6 weeks after conception and is also characterized by the *fusion of the tracheoesophageal folds* to form a *ventral laryngotracheal tube* and a *dorsal esophagus*.

This process is usually impaired in children with a tracheoesophageal fistula as the
Clinical Presentation of Congenital Tracheoesophageal Fistulas

*Esophageal atresia*, a key component in most types of tracheoesophageal fistulas, can be diagnosed prenatally. *Maternal polyhydramnios* is usually evident in most cases and *prenatal ultrasonography* can reveal stomach’s gas absence.

During the neonatal period, the most common presentation is the development of *copious amounts of white frothy bubbles* in the mouth and nose. These bubbles recur despite repeated suctioning. Infants with congenital tracheoesophageal fistulas usually present with *respiratory distress symptoms* and *recurrent pneumonia*.

*Coughing*, *choking* and *cyanosis* can be seen in an infant with a congenital tracheoesophageal fistula especially during feeding.

Most TEF present almost immediately after birth.

- Food cannot get to stomach, resulting in *immediate emesis* and *rapid dehydration*.
- Gastric contents may enter lungs, resulting in *respiratory distress*.

The main goal of physical examination in an infant with a tracheoesophageal fistula is to exclude any of the previously mentioned anomalies that can be associated with this congenital malformation.

Diagnostic Workup for Congenital Tracheoesophageal Fistulas

Laboratory investigations are not useful in establishing the diagnosis of a congenital tracheoesophageal fistula. *Prenatal and postnatal imaging studies* are the main diagnostic tools to establish the diagnosis of a tracheoesophageal fistula.

*Prenatal ultrasonography* can reveal *polyhydramnios, absence of gas in the stomach, a distended esophageal pouch* and *in-utero growth retardation*. These findings are indicative of esophageal atresia and not a tracheoesophageal fistula per se, but because esophageal atresia is seen in most cases of tracheoesophageal fistulas, establishing the diagnosis on the confirmation of an esophageal atresia is reasonable.
Neonates or infants with a congenital tracheoesophageal fistula should undergo chest x-ray imaging to exclude complications such as aspiration pneumonia and to have more clues to confirm the diagnosis. The most common findings on a chest x-ray are tracheal compression and deviation, or absence of a gastric bubble.

A nasogastric tube might be inserted to confirm the diagnosis of esophageal atresia. When esophageal atresia is present, the nasogastric tube will coil in the mediastinum.

Multidetector-row computed tomography scans can be used to confirm the diagnosis of a tracheoesophageal fistula without the use of a contrast medium. Because of the non-invasiveness of the procedure, computed tomography scans become preferable by most pediatric surgeons to plan the repair procedure for tracheoesophageal fistulas.

**Treatment of Congenital Tracheoesophageal Fistulas**

It is essential to transfer any neonate or infant who is diagnosed with a tracheoesophageal fistula to a pediatric hospital that has experience in the surgical repair of tracheoesophageal fistulas. The early repair of the fistula has been associated with excellent survival rate.

Surgical repair can be delayed in neonates with very low birth weight or aspiration pneumonia. All other healthy infants should be provided primary surgical repair of the fistula within the first few days of life. A cuffed endotracheal tube should be placed distal to the fistula to prevent aspiration during the operation.

Once the fistula is repaired, care must be taken to prevent gastroesophageal reflux and prolonged mechanical ventilation as they were associated with an increased risk of recurrent tracheoesophageal fistulas. Use of a tracheostomy, elevation of the head of the bed and frequent suctioning of oral secretions are indicated to prevent recurrence.
References


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