Congenital Tracheoesophageal Anomalies

Developed by Shawn Dodd and Dr. Bryan Dicken for PedsCases.com.
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Introduction:

Hello listeners, my name is Shawn Dodd and I am a third-year medical student at the University of Alberta. Today's podcast was developed with the help of Dr. Bryan Dicken, a pediatric surgeon at the University of Alberta and is designed to give you an understanding of congenital tracheoesophageal malformations. As congenital tracheoesophageal malformations require early surgical management it is important to recognize presenting features.

So, to get us in the clinical mindset, let's start off with a case.

Case Presentation:

You are a medical student on your General Surgery rotation with the Pediatric Surgery Team. Your team gets called to see a neonate with excessive salivation at birth. On arrival you are presented with a male neonate weighing 3160g, delivered via spontaneous vaginal delivery at 39 weeks and three days. You are told that the mother's pregnancy was unremarkable. At delivery, the infant required no resuscitation and had Apgar scores of 9 at both 1 and 5 minutes. As baby and Mom were coping well post-delivery, the infant was placed skin-to-skin on Mom's chest for one hour. One hour after delivery, the infant's physical exam was remarkable for white, frothy oral secretions which were eliminated with suctioning.

It is now 5 hours into the infant's life and the healthcare team is concerned as the infant continues to have excessive salivation and difficulty feeding. Mom states that during his first time feeding, the infant began coughing and choking. Moreover, the nursing team noticed that the infant's abdomen appears quite distended.

Your resident turns to you and says "This sounds like a possible tracheoesophageal malformation. What type of tracheoesophageal malformation does this sound like to you? What should we do next?"

***Objectives*:**

To help you answer these questions, this podcast has three main objectives:

1) Review the anatomical differences between the various types of tracheoesophageal anomalies;
2) Develop an approach to an infant presenting with a tracheoesophageal anomaly;
3) Briefly review the surgical approaches used to repair tracheoesophageal anomalies.

What are tracheoesophageal malformations?

Let's start with the fundamental question, what are tracheoesophageal malformations? Tracheoesophageal malformations can be divided into two main types of anatomical abnormalities: 1) tracheoesophageal fistulas, and 2) esophageal atresia. Tracheoesophageal fistulas are an abnormal connection between the trachea and the esophagus (Kovesi & Rubin, 2004). Though tracheoesophageal fistulas can be acquired through causes such as cuff-related injuries during tracheal intubation, trauma, infection or ingestion of foreign bodies, such as a battery (Santosham, 2018), this podcast will focus on congenital tracheoesophageal anomalies. Though congenital tracheoesophageal fistulas can occur without any additional abnormalities, it is more common for them to present concurrently with esophageal atresia (UpToDate, 2019). In fact, it is estimated that the incidence of esophageal atresia is 1.7 in 10,000 births, with over 90% of these children having an associated tracheoesophageal fistula (Kliegman *et al.,* 2016)

This bring us to the second type of tracheoesophageal anomalies: esophageal atresia. Esophageal atresia refers to the complete interruption in the continuity of the esophageal lumen (Kovesi & Rubin, 2004). Such an interruption leads to two incomplete segments of the esophagus. The proximal segment starts in the oropharynx and travels down into the mediastinum. The distal portion originates in the mediastinum and terminates in the stomach. As with tracheoesophageal fistulas, esophageal atresia also rarely occurs in isolation. Anatomical variations of congenital tracheoesophageal malformations are classified using the Gross classification system (DynaMed, 2019).

The Gross classification systems classifies tracheoesophageal fistulas and esophageal atresia into 5 types (DynaMed, 2019). Type A describes an isolated esophageal atresia where the proximal and distal portions of the esophagus are not continuous with each other. In Type A, no tracheoesophageal fistulas exist. Type B is when the proximal and distal esophagus are not continuous, and the proximal portion of the esophagus has fistulized into the trachea. Type C is similar to type B, but in this type the distal esophagus has fistulized with the trachea. Type D occurs when both the proximal and

distal portions of the esophagus fistulize with the trachea forming two separate fistulas. Lastly, Type E occurs when there is no esophageal atresia, but a fistula exists between the lumen of the esophagus and trachea. Type E tracheoesophageal fistulas are also referred to as H-type fistulas as their appearance resembles a capital H.

Clinical Presentation

Prenatal Indications of Tracheoesophageal Malformations

Now that we understand what tracheoesophageal malformations are and how to classify them, let's review clinical presentation. While most tracheoesophageal malformations are diagnosed shortly after delivery, some can be recognized prenatally. Risk factors associated with tracheoesophageal malformations include advanced maternal age, obesity, European descent, low SES and tobacco consumption (Kliegman *et al.,* 2016). Early signs of a potential tracheoesophageal malformation may be identified on prenatal ultrasound as early as 24 weeks gestation (Pardy *et al.,* 2019). Suggestive ultrasound findings include: polyhydramnios, absence of a fluid-filled stomach, small abdomen, a distended esophageal pouch, or anechoic area in middle of fetal neck. These findings are consistent with a proximal GI obstruction and appear due to a failure of the fetus to swallow and absorb amniotic fluid. In patients these findings, a high-resolution fetal ultrasound or fetal magnetic resonance imaging may be warranted (Dynamed, 2019; Slater & Rotherberg, 2016).

Postnatal Indications of Tracheoesophageal Malformations

Most infants with a tracheoesophageal malformation are symptomatic in the first few hours of life. Typically, these present as excessive bubbling or frothing at the mouth and/or nose. Feeding difficulties are noted early with regurgitation, gagging, coughing, choking, and possible respiratory distress with feeds. Alternatively, malformation may be recognized when a suction catheter, or a naso/orogastric tube cannot be advanced.

In patients with a tracheoesophageal fistula without esophageal atresia, the clinical symptoms may be less apparent, and therefore these patients may have delayed presentations (Karnak *et al.,* 1997; Suen, 2018). Typically, the presentation is more ambiguous and can include recurrent aspirations, multiple episodes of pneumonia, abdominal distension and a history of cyanosis during feeding. As symptoms are less acute than patients with esophageal atresia, more invasive means may be required to reach a diagnosis (Karnak *et al.,* 1997). We will further explore these diagnostic tests later in this episode.

Physical Exam

As with all newborn infants, a thorough head-to-toe examination should be done at birth and prior to discharge from hospital. This is especially important in infants with tracheoesophageal malformations as up to half of these infants have other congenital

anomalies. Approximately 19% (DynaMed, 2019) of cases are associated with either VACTERL or CHARGE sequences. VACTERL sequence refers to patients presenting with vertebral anomalies, anorectal anomalies, cardiac malformations, tracheoesophageal fistula, renal anomalies and radial limb anomalies. CHARGE sequence refers to patients with coloboma, heart defects, atresia of the choanae, mental and/or physical development delays, genital hypoplasia, and ear abnormalities.

Investigations

The investigations needed to diagnose tracheoesophageal malformations differ based on the presence of esophageal atresia. In patients with suspected esophageal atresia, a diagnosis can be made by attempting to pass a catheter into the stomach (Clark, 1999; UpToDate, 2019). If a catheter cannot be passed into the stomach, a chest x-ray can be performed. If the catheter can be visualized curled up in the upper portion of the esophagus, esophageal atresia can be diagnosed. Very rarely, a small amount of water-soluble contrast can be placed into the upper portion of the esophagus to confirm the presence of esophageal atresia. This test is rarely performed, however, as accidental aspiration of the contrast fluid may result in pneumonitis.

Isolated tracheoesophageal fistulas without esophageal atresia is more challenging to diagnose. The gold-standard for diagnosis of an H-type fistula is a prone esophogram (Ke *et al.,* 2015; Suen, 2018). Alternately, bronchoscopy and esophagoscopy may be employed.

All patients should also have an echocardiogram to evaluate the structure and function of the heart, and most importantly, to determine the location of the aortic arch. Given that approximately 5% of patient will have a right-sided arch, it is critical to be aware of the location of the aortic arch in order to determine which side to approach during surgery. Cranial and abdominal imaging may be required to rule out other associated abnormalities.

Finally, given that 19% tracheoesophageal malformation cases are associated with either the VACTERL or CHARGE sequences, consultation of Pediatric Urology, Cardiology, Otolaryngology, Ophthalmology, Pulmonology, Orthopedic Surgery and, Clinical Genetics and Genomics is warranted if additional anomalies are apparent or of concern. Moreover, a Clinical Dietician should be consulted to assist with nutritional support and a Speech Language Pathologist should be involved post-operatively to support feeding.

Short-term Management

As you may suspect, tracheoesophageal malformations with esophageal atresia require a different short-term management compared to isolated tracheoesophageal fistulas. In patients with esophageal atresia a Replogle tube with continuous suction should be inserted in the proximal portion of the esophagus (Clark, 1999) and oral feeding should

be stopped. This, along with elevating the patient head and neck prevents the collection of secretions and decreases the risk of aspiration.

If patients begin to experience respiratory distress, bag masks should not be used. In patients with tracheoesophageal fistulas, bag masks may result in accidental insufflation of the stomach resulting in acute gastric distension or rupture. If endotracheal intubation is required, care must be taken to ensure the cuff is below the fistula to avoid insufflation of the GI tract, which may result in acute respiratory failure due to elevation of the diaphragm preventing adequate ventilation. As such, fiber-optic bronchoscopy should be considered to ensure the cuff lies below the fistula. Finally, General Surgery should be consulted. If surgery is delayed, a gastrostomy tube should be considered to enable gastric venting.

Surgical Management

Now that the patient is stabilized, the final step in management is surgery. When planning a surgical repair of a tracheoesophageal malformation we need to consider how the repair will be approached and which malformations need to be repaired.

Two main surgical approaches exist: 1) thoracotomy with an extrapleural approach and 2) thoracoscopic approach (Slater, 2016). For both approaches, the patient should be positioned in a left lateral decubitus position. This position improves access to the posterior mediastinum and enables the thorax to be accessed from the patient's right side, as the left side contain the heart and great vessels. In a thoracotomy with an extrapleural approach, the thorax is entered through the fourth intercoastal space with attention taken to avoid opening the pleura. By not opening the pleura, any post-operative leaks will not drain into the pleural cavity and risk causing an empyema (Slater, 2016).

In the thoracoscopic approach, three ports are used to access the malformation. With the thoracoscopic approach, the pleura is opened and the procedure is performed within the pleural cavity (Slater, 2016). Though this approach does decrease morbidity as it does not require the large incision associated with the thoracotomy approach, there are contraindications to the thoracoscopic approach (Slater, 2016). Relative contraindications include significant cardiac defects, prematurity and small size (<1500 g), and significant abdominal distension. Hemodynamic instability is an absolute indication.

Now that we've planned how we will access the malformation, we next need to consider what actually needs to be repaired. The first consideration that needs to be made is whether or not a tracheoesophageal fistula exists. If a fistula is present, the primary objective of the operation needs to be to dissect and ligate all fistulas to prevent airway complications. Once all the tracheoesophageal fistulas have been identified, dissected and ligated, the presence of esophageal atresia must be addressed.

For esophageal atresia, the type of repair that will be performed will depend on the distance between the proximal and distal segments of the esophagus. If the gap between the proximal and distal segments is short, a primary anastomosis can be performed (Dynamed, 2019). If the gap between the proximal and distal portion is long, however, four main strategies exist to anastomose the esophagus. The preferred method is a delayed primary repair, where the infant is stabilized and a Replogle tube is place into the proximal esophagus with continuous suction (Friedmacher & Puri, 2012). A gastrostomy tube is then placed to support feeding and gastric venting. The infant is then monitored for 12 weeks, after which point a gap study is performed to evaluate the distance between the two ends. If this is favorable, a primary anastomosis of the esophagus is attempted. As it is accepted that conservation of the native esophagus yields the best outcomes, other techniques may also be employed to encourage the proximal and distal segments to elongate. Such techniques include the Foker procedure where sutures are placed at the end of the esophagus segments and then brought externally to the skin's surface. Tension is then applied to the suture causing mechanical stretch to the esophageal segments. This method is thought to increase the rate of growth of the segments. Once the segments are of adequate length, another operation is performed to anastomose the esophagus.

In certain patients, the native esophagus cannot be conserved due to the long gap. In these patients two techniques can be performed: 1) gastric transposition and 2) colonic interposition. As their names suggest, gastric transposition involves the stomach being pulled superiorly and connected to the proximal esophageal segment. In colonic interposition, a segment of colon is isolated and relocated into the thorax to connect the proximal and distal segments of the esophagus (Dynamed, 2019).

Complications:

Although the survival rate of patients with tracheoesophageal malformations has greatly improved in recent history, these patients remain at risk for long-term complications. As aforementioned, tracheoesophageal malformations are often associated with VACTERL and CHARGE syndrome, whose many anomalies creates a medically complex patient (Clark, 1999; DynaMed, 2019; UpToDate, 2019). Moreover, after repair most patients with esophageal atresia will generally experience some degree of esophageal dysmotility, dysphagia and gastroesophageal reflux disease and may require proton-pump inhibitor therapy for at least one year. In addition to the typical risks associated with pediatric surgery, patients with repaired tracheoesophageal malformations are also at risk of developing strictures in either the esophagus or the trachea, especially at the site of anastomosis, and possible re-fistulisation. Unfortunately, the complications associated with the diagnosis of a tracheoesophageal malformation also continue into childhood and adolescence (Krishnan *et al.,* 2016). These patients remain at an increased risk of developing peptic esophagitis, Barrett's esophagus and gastric metaplasia and feeding disorders. As such, these patients require long-term follow-up with a Pediatric Gastroenterologist to ensure appropriate management of feeding and nutrition and ensure appropriate preventative screening is provided.

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Case Conclusion:

So, let's go back to our case. Your resident just asked you, "What type of tracheoesophageal malformation does this sound like to you? What should we do next?"

Given that your patient has excessive salivation and difficulty feeding, you can suspect that some degree of esophageal atresia exists. Moreover, given that the infant's abdomen is distended, it is possible that there is a fistula between the distal esophagus and trachea. As such, you suspect that the infant has either a Type C or D tracheoesophageal fistula based on the Gross Classification.

To support your patient, you propose inserting a Replogle tube with continuous suction in the proximal portion of the esophagus and positioning the infant with their head slightly elevated to manage secretions and decrease the risk of aspiration. Next, you order a chest x-ray to confirm your suspicions, an echo to assess the structure and function of the heart and its vessels and an umbilical catheter for IV access. Finally, you book an OR for surgical repair of the malformation and suggest the patient be transferred to the NICU for monitoring during the pre- and post-operative period.

Now that you've stabilized the patient we want to leave you with a few last summary points regarding tracheoesophageal malformations.

1. Tracheoesophageal malformations can be divided into two main types of anatomical abnormalities: tracheoesophageal fistulas, and esophageal atresia. A tracheoesophageal fistula is an abnormal connection between the trachea and the esophagus. Esophageal atresia refers to the complete interruption in the continuity of the esophageal lumen. These two abnormalities often present concurrently. There are 5 types of tracheoesophageal malformations which can be classified per the Gross Classification system.

2. Tracheoesophageal malformations typically present with feeding difficulties and respiratory distress in the neonatal period. In patients with an isolated tracheoesophageal fistula, the clinical symptoms may be less apparent leading to a delay in presentation.

3. If you suspect a tracheoesophageal malformation, don't forget to do a head-to-toe assessment of the newborn. Approximately 19% of tracheoesophageal malformations cases are associated with either the VACTERL or CHARGE sequences.

4. Short term management of a suspected tracheoesophageal malformation includes stopping oral feeds, inserting a Replogle tube with continuous suction, and positioning the infant with their head slightly elevated to manage secretions. The Pediatric Surgery team should be promptly consulted for surgical management.

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5. Though the survival of patients with tracheoesophageal malformations has greatly improved, these patients remain at risk for many long-term complications. A multidisciplinary team should be involved in ongoing patient care.

And with that, we thank you for listening and we hope you enjoyed this podcast! Best of luck!

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