Oesophageal atresia and tracheo-oesophageal fistula

Spencer W Beasley

Abstract

Oesophageal atresia and tracheo-oesophageal fistula is a congenital structural abnormality that affects 1:4500 live infants. It is due to failure of the primitive foregut tube to separate correctly into oesophagus and trachea. About 50% have associated abnormalities, of which the VACTERL (Vertebral, Anorectal, Cardiac, Tracheo-oEsophageal, Renal and Limb) association is the most common. Prematurity is common and all have some degree of tracheomalacia. Surgery of the common type can be performed through a fourth interspace thoracotomy or by thoracoscopy. It involves division of the distal tracheo-oesophageal fistula and anastomosing together the two ends of the oesophagus. The absence of a distal fistula reveals itself as a 'gasless abdomen' on plain radiology, and usually indicates a long gap between the blind oesophageal ends: this sometimes necessitates an oesophageal replacement if extensive oesophageal mobilization fails to achieve an end-to-end anastomosis of the oesophagus. Potential postoperative problems include anastomotic leak, anastomotic stricture, recurrence of the fistula, gastro-oesophageal reflux, oesophageal dysmotility and food impaction. Survival is determined mainly by coexisting congenital abnormalities. The long-term risk of oesophageal malignancy is yet to be established. Isolated tracheo-oesophageal fistula ('H fistula') can occur without atresia, and often presents after feeding has commenced. It is divided through a cervical incision.

Keywords Anastomosis; gastro-oesophageal reflux; oesophageal atresia; oesophageal replacement; prematurity; tracheo-oesophageal fistula; tracheomalacia; VACTERL association

Background

Oesophageal atresia (OA) is a congenital structural abnormality of the foregut that results in a variable length of the mid-portion of the oesophagus to be missing. Typically, there is an abnormal attachment of the lower oesophageal segment to the trachea, called a distal tracheo-oesophageal fistula (TOF). Much less commonly there is a proximal TOF, which is an abnormal connection between the upper oesophageal segment and the trachea. Even more rarely, both proximal distal fistulae are present.

OA with or without TOF, should be diagnosed before or at birth, prior to the first feed being given. In OA, food entering the blind upper oesophageal segment has nowhere to go – except back up, or into the trachea which then spills into the lungs.

This article provides an overview of the diagnosis and management of OA and its implications beyond the neonatal period.

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Pathoembryology

Early in gestation, the developing embryo has a single foregut tube. Lung buds sprout from its ventral aspect. Shortly afterwards, and proximal to this, the tube divides into two parallel tubes: the anterior tube becomes the trachea, and the posterior one becomes the oesophagus. This process is orchestrated by the notochord which influences the expression of the Sonic Hedgehog gene, a process which is under strict temporo-spatial control. Any abnormality in the timing or location of this expression leads to abnormalities of tracheo-oesophageal division, as well as other abnormalities seen as part of the VACTERL (Vertebral, Anorectal, Cardiac, Tracheo-oEsophageal, Renal and Limb) association. It is accepted that aberrations in the normal process of separation and growth of the oesophagus and trachea may lead to OA and TOF. The process involved is complex and has not been fully elucidated, but Nkx2.1, Tbx4, FGF10, Gli factors, and SHH have now all been implicated in the pathogenesis of oesophageal atresia.

Classification

The variety of different anatomical configurations of OA and TOF have led to a number of classification systems, but the most widely used is shown in Figure 1. By far the most common type (about 85%) is OA with a distal TOF, termed a type C in the Gross classification. The most difficult to treat are types A and B, where there is no distal fistula and the gap between the blind oesophageal ends tends to be substantial, so-called 'long gap oesophageal atresia'. Due to the wide range of variations in OA, many surgeons prefer to avoid any confusion by being specific in their description of each case rather than by adhering to one of the classifications, e.g. pure OA with a long gap etc.

While not strictly OA at all, an isolated tracheo-oesophageal fistula ('H-fistula') is usually included in discussion of OA (see Management of Specific Situations below).

Incidence

Although there are some geographical variations in incidence of OA, the overall incidence is believed to be about 1:4500 live births. It occurs in all regions and all ethnic groups. The recurrence risk for siblings of a child with isolated OA/TOF is less than 1%.

Antenatal diagnosis

Sometimes OA can be diagnosed on antenatal ultrasonography. Clues to its existence include a dilated upper oesophagus, small or absent stomach, abnormal swallowing, maternal polyhydramnios, and recognition of other abnormalities that are known to coexist with OA, such as those of the VACTERL association. These features raise the suspicion of OA, such that as soon as the baby is born, the definitive diagnosis still needs to be confirmed.

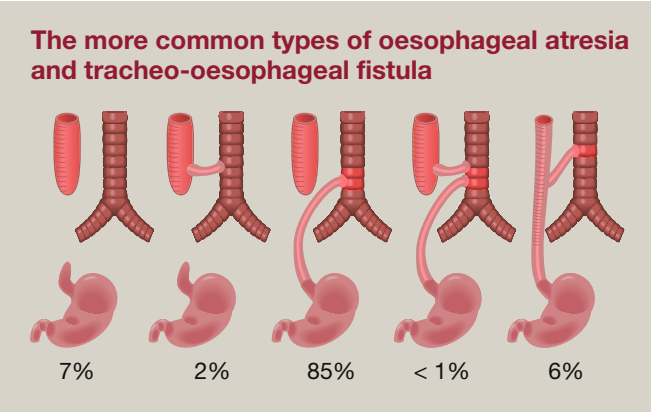

Figure 1

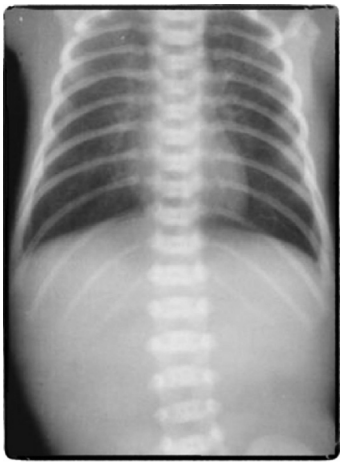

Figure 2 The gasless abdomen signifies absence of a distal tracheo-oesophageal fistula.

Clinical presentation

The classical feature at birth is of an excessively mucousy drooling infant that results from excessive saliva accumulating in the blind upper oesophageal pouch. Any infant with these features should not be fed until the diagnosis of OA has been excluded. Other clinical signs include tachypnoea, coughing, and choking.

About one-third of infants with OA are born prematurely and about 30% have a history of maternal polyhydramnios: this percentage is much higher in those without a distal fistula. Where there are other abnormalities that have a known association with OA, the possibility of OA must always be considered. For example, if there has been polyhydramnios, or if there is an anorectal malformation, the patency of the oesophagus must be confirmed by passing an orogastric tube through it shortly after birth.

Investigations

There are three stages in the investigation of every infant with suspected OA.

1. *Confirm atresia of the oesophagus.* This is done by passing an 9–10F orogastric tube through the mouth into the oesophagus. In OA such a tube cannot be introduced further than 9–10 cm from the gums. This is the only test required to make the diagnosis. A finer tube is not used because it can curl around in the upper oesophageal pouch giving a false impression of oesophageal continuity.
2. *Determine the type of atresia by establishing whether there is a tracheo-oesophageal fistula*. The presence of a distal TOF is done by taking a plain x-ray of the torso: air below the diaphragm, within the bowel, confirms that there must be a connection between the trachea and stomach, i.e. a distal tracheo-oesophageal fistula. No gas below the level of the diaphragm implies no distal fistula (Figure 2). The further investigation of an infant with a 'gasless abdomen' is described later.
3. *Identify any coexistent abnormalities that may influence the management of the OA*. The key abnormalities are those of the VACTERL association (Vertebral, Anorectal, Cardiac, Tracheo-oEsophageal, Renal and Limb), CHARGE association and major chromosomal abnormalities.

Most significant congenital heart disease is now diagnosed on antenatal ultrasonography, and the passage of urine after birth signifies functioning kidneys. However, most centres still perform echocardiography and obtain a renal ultrasound scan. Examination of the perineum will diagnose an anorectal malformation, and the thoraco-abdominal plain X-ray (already performed to diagnose a distal fistula) also allows assessment of the vertebrae and ribs. Radial and thumb abnormalities are evident on clinical examination. Sometimes these abnormalities influence the immediate management, including the timing and type of surgery. For example, duct-dependent congenital heart disease may require immediate commencement of a prostaglandin E infusion, and surgery to the OA is deferred until the infant is stable. If there are multiple gastrointestinal abnormalities, the correct order of repair is from proximal to distal. This means that closure of the fistula and repair of the oesophagus precedes duodeno-duodenostomy for duodenal atresia, and finally a colostomy for a concomitant high anorectal malformation.

Major chromosomal aberrations occur in about 7% of cases of OA/TOF. The most important of these are trisomy 13, 18 and 21. If suspected, urgent karyotyping is performed because, for example, if trisomy 18 is present, definitive surgery for the OA may not be appropriate.

Initial treatment

The normal care and monitoring of the surgical newborn is initiated. Specific attention is paid to maintaining normothermia at a time where multiple other investigations and interventions are taking place.

In addition to this, it is essential that continual or frequent aspiration of the saliva accumulating in the upper oesophageal pouch is initiated to prevent aspiration. This is achieved by insertion of a suction catheter with frequent aspiration of saliva, or positioning of a Replogle tube with continuous suction. This continues until the definitive operative repair has been performed.

The parents should be carefully informed throughout, updating them about what is planned, including details of the purpose and likely outcomes of all procedures. They will need to provide informed consent for the operation.

Surgery

The definitive operation (Figure 3) involves:

- division and closure of the tracheo-oesophageal fistula; and
- joining the two blind ends of the oesophagus together (end-to-end oesophageal anastomosis).

This can be achieved either through a 4th intercostal space extrapleural thoracotomy or by a transpleural thoracoscopic approach. The thoracotomy incision is transverse, and centred on the inferior angle of the scapula. The fibres of latissimus dorsi can be divided in line with the incision, but the serratus anterior should be left intact and retracted (to avoid a winged scapula from damage to the long thoracic nerve). Once in the chest, irrespective of the surgical approach, the azygos vein is divided and the tracheo-oesophageal fistula identified at the point at which it enters the trachea. It is divided and closed. The two ends of the oesophagus are mobilized sufficiently to enable an end-to-end all-layered anastomosis without excessive tension. A chest drain is not routinely required and may increase the anastomotic leakage rate, but may have a place where the anastomosis is marginal. Some surgeons routinely use a trans-anastomotic tube, but the author only uses this for premature infants who are predicted to need gavage feeding, in which situation a small calibre gavage feeding tube is used.

Oral feeds (preferably colostrum and breast milk) are commenced from about 48 hours. The uncomplicated full-term infant can usually be discharged from hospital at about 5 days. Most surgeons perform a postoperative contrast study at some stage, either early to rule out an anastomotic leak prior to commencing feeds, or after discharge from hospital to assess the anastomosis, oesophageal function, and gastro-oesophageal reflux.

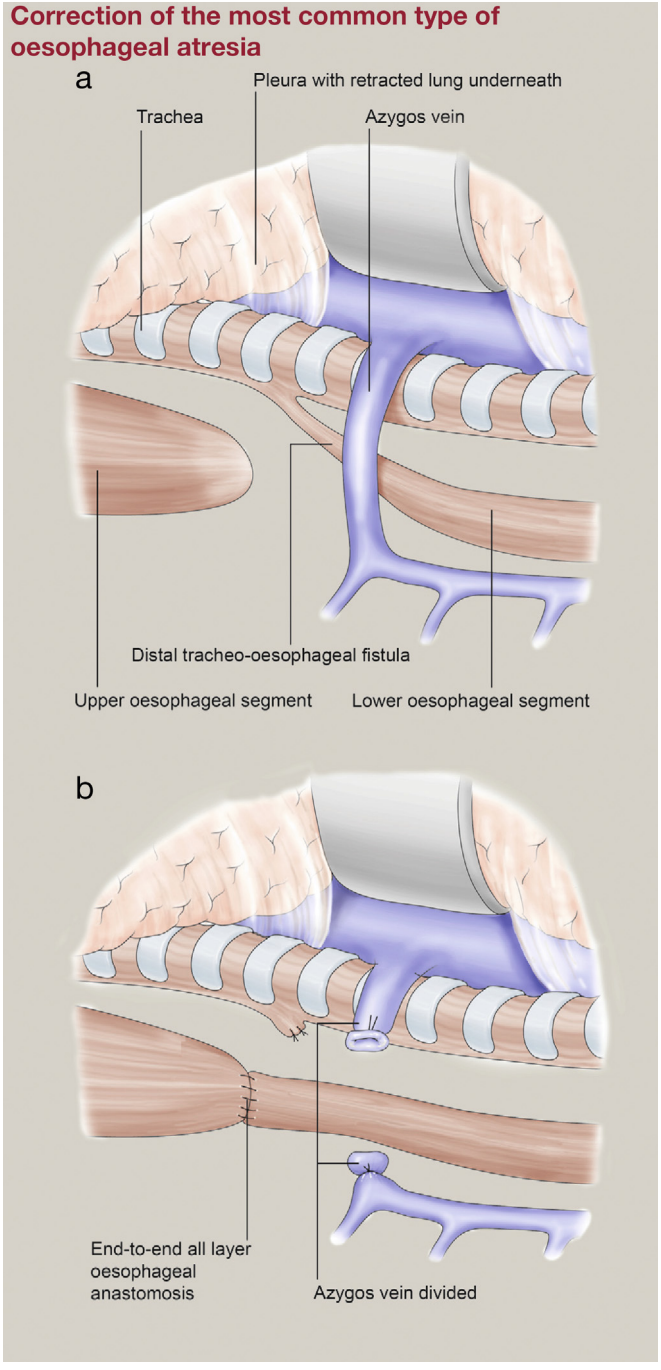

Figure 3 Surgery involves division and closure of the distal tracheo-oesophageal fistula and mobilization of the oesophageal ends to allow an end-to-end anastomosis.

Management of specific scenarios

Extreme prematurity

Infants with OA born extremely prematurely present additional problems: not only are they very small, and their immature tissues provide the surgeon with additional technical challenges, but also their lungs are immature and they are at risk of hyaline membrane disease (HMD). The risk is that there will be preferential passage of air through the distal fistula as the HMD worsens, and this may lead to gastric dilatation which worsens the situation further by causing elevation and splinting of the diaphragm — impeding ventilation that is already compromised. Prior to the availability of surfactant therapy, the eventual outcome was of increasing respiratory difficulty, gastric perforation, tension pneumoperitoneum and death unless the fistula could be controlled promptly. If there is tension pneumothorax, this is best managed by urgent release of the tension by needle paracentesis followed by immediate laparotomy, with introduction of a Foley catheter through the gastric perforation (which is usually on the anterior surface of the stomach) into the lower oesophagus where it tamponades the oesophagus. This allows formal closure of the fistula by thoracotomy.

The gasless abdomen

A gasless abdomen signifies that there is no distal TOF. However, in about 20% there is a proximal fistula. This can be identified by performing a contrast study of the upper oesophagus under fluoroscopic control looking for passage of contrast through the fistula into the airways, but runs the risk of contamination of the airway if there is a fistula present. Alternatively, and more reliably, the upper pouch fistula can be seen on bronchoscopy as it enters the posterior wall of the upper trachea. A proximal fistula is repaired at the time of definitive repair of the oesophagus, or earlier if it is creating difficulties.

The main problem in OA without a distal fistula is that the gap between the oesophageal ends tends to be long. Often the distal oesophageal segment is short, only just extending above the level of the diaphragm. The extent of gap between the ends influences the timing and type of definitive repair. Consequently, a priority is to measure the gap: this is done at the time of initial gastrostomy (which is required to establish early enteral feeding) by introducing a metal sound through the stomach and through the gastro-oesophageal junction into the lower oesophagus at the same time as the anaesthetist introduces a radio-opaque tipped catheter into the upper oesophagus. Fluoroscopy is performed on the table to assess the gap between the two.

If the gap is more than 4 vertebral bodies (suggesting that an oesophageal anastomosis is likely to be difficult or impossible) definitive surgery is delayed until about 3 months. With time the oesophageal ends seem to lengthen slightly, and certainly the tissues gain strength and become more able to sustain an anastomosis under tension. On the other hand, if the gap is smaller, and it is felt that extensive mobilization will enable the ends to be brought together, definitive repair is performed earlier.

A summary of the management of OA where there is no distal fistula is given in Box 1.

Tracheo-oesophageal fistula without atresia, the 'H fistula'

Although the oesophagus is intact and in continuity, for various reasons this condition tends to be included in any discussion of OA. It tends not to be diagnosed before birth and may not be obvious even at birth. Because the oesophagus itself is intact, the infant can swallow, but at some stage milk (or other feeds) may pass from the oesophagus into the airway causing choking, spluttering. or cyanosis. Alternatively, air may pass in the reverse direction through the fistula causing abdominal distension. The diagnosis is confirmed on bronchoscopy or a prone mid-oesophageal contrast study. It is normally repaired through a right cervical approach taking great care to avoid damage to the recurrent laryngeal nerves. The fistula is best divided surgically (rather than by ligation alone) to minimize the risk of a recurrent fistula developing. Various techniques for the endoscopic occlusion of the fistula have also been described, but numbers are small, and it is difficult to judge their efficacy.

Complications and long-term issues

Anastomotic leakage

This occurs as a result of either technical error during construction of the anastomosis, or due to relative ischaemia of the oesophageal ends resulting from excessive mobilization or excessive tension (paradoxically from insufficient mobilization). It is rare after repair of OA with a distal TOF, but is more likely with long gap atresia. If it is asymptomatic, such as observed during a postoperative contrast study, it can be treated conservatively and it will usually resolve spontaneously. At the other end of the spectrum, however, complete anastomotic dehiscence will require further surgery. Other leaks can be expected to settle with time, but require cessation of oral feeds, introduction of TPN, and treatment with broad-spectrum antibiotics until they have healed.

Summary of the management of OA with a gasless abdomen

1. Establish whether there is a proximal TOF
   - Upper pouch contrast study under continuous fluoroscopic control
   - Bronchoscopy to directly visualize the fistula
2. Initial gastrostomy
   - Open procedure
   - Allows enteral feeding to be established early
3. Determine the length of the gap between the two blind-ending oesophageal segments
   - Done at time of gastrostomy
   - Anaesthetist inserts radio-opaque tipped catheter into upper pouch
   - Surgeon inserts metal sound through gastrotomy into lower oesophagus
   - Distance between both assessed on Xray
4. Decision on timing and type of definitive repair
   - According to length of gap, surgical expertise, concomitant congenital abnormalities/prematurity
5. Ongoing upper pouch suction until repair conducted
   - Avoids aspiration of saliva and choking episodes
6. Definitive repair using oesophagus
   - Thoracotomy/thoracoscopy
   - Full mobilization of upper oesophageal segment
   - Identification and mobilization of lower oesophagus to diaphragm
   - Avoid damage to the vagus
   - Myotomies rarely performed now because of their high complication rate: ischaemia, stricture, leakage
   - Foker manoeuvre an alternative in some centres with variable results
7. Oesophageal replacement
   - Inability to approximate the oesophageal ends is an indication for oesophageal replacement
   - Options include:
     - proximal or distally based greater curvature gastric tube,
     - Scharli procedure,
     - gastric transposition,
     - colonic interposition, or
     - jejunal interposition.
   - Most centres now use stomach for replacement, but each procedure has its advocates

Box 1

Anastomotic stenosis/stricture

Strictures result from technical error, ischaemia, or excessive tension at the anastomosis, and can be exacerbated by gastro-oesophageal reflux (GOR). Some minor narrowing on the contrast study is common, and if asymptomatic does not require

treatment. More severe narrowing causes symptoms, with dilatation of the upper segment and delay in the passage of food and fluid through the stricture. A severe stricture requires dilatation which is usually done by radial balloon dilatation under fluoroscopic control. Coexisting GOR is managed by commencing a proton pump inhibitor, which reduces the likelihood of recurrence of the stricture. Sometimes multiple dilatations are required.

Recurrent fistula

These occur rarely now that the fistula is divided rather than ligated, and now that the fistula is closed with absorbable sutures rather than with silk. Should a fistula recur, it must be closed once the infant is stable, once sepsis has been controlled, and once the infant is anabolic and receiving good nutrition. The fistula is normally closed surgically, but various other techniques have been described as well.

Tracheomalacia

This is invariably present in all cases of OA, but varies considerably in severity. The collapse of the trachea, particularly during expiration, causes difficulty in breathing. The natural history is for improvement with time, as the infant grows. Occasionally it is so severe that it demands surgery to support the trachea to help keep it open (tracheopexy or aortopexy).

Gastro-oesophageal reflux

This is common in OA, and is probably exacerbated by mobilization of the lower oesophagus during repair of the atresia. As outlined above, it has the potential to cause the anastomosis to stricture. The situation is made worse because the inherently poor peristalsis of the oesophagus in OA means that any reflux of gastric contents into the oesophagus clears more slowly than normal, lengthening the period of exposure of the anastomosis and mucosa to the damaging effects of acid. It is better to treat the reflux with anti-acid medication rather than surgery because a fundoplication is a fine balance between stopping the reflux and worsening swallowing if the fundoplication itself makes oesophageal clearance even more difficult. Some surgeons perform a 'partial wrap' fundoplication in these circumstances.

Food impaction and dysphagia

Dysphagia is a direct consequence of the poor oesophageal function inherent in OA, and sometimes is exacerbated by vagal damage during repair. It tends to improve with time, but most children and adults with repaired OA will take a glass of water with their meals and have to be careful to chew their food properly. Food impaction is common between the ages of 1–4 years. If the food bolus does not dislodge spontaneously (fizzy drinks can sometimes aid this), endoscopic removal is needed. At the time of oesophagoscopy the anastomosis should be reviewed in case a stricture at the anastomosis has been a contributing factor.

Risk of malignancy

Previously this was thought to represent a significant long-term risk in these patients, but the current evidence suggests that oesophageal malignancy is only slightly more common than in the general population, but it occurs at a younger age. There remains uncertainty as to whether regular surveillance of the oesophagus is required in all patients outside the research setting. It seems prudent to do so in patients known to have long-standing gastro-oesophageal reflux.

Survival

The main determinant of survival is the presence or absence of severe co-existing abnormalities, e.g. complex congenital heart disease, bilateral renal agenesis and trisomy 18. It is now rare that the complications of the OA itself, or its treatment, cause mortality. ◆

FURTHER READING

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