Oesophageal Atresia

Oesophageal atresia is a congenital abnormality in which there is a blind ending oesophagus. It can occur in isolation or there may be one or more fistulae communicating between the abnormal oesophagus and the trachea, known as a tracheo-oesophageal fistula (TOF).[^1] The exact aetiology is uncertain but there appears to be a defect in embryological development. Various hypotheses have been put forward suggesting possible genetic causation and teratogenic influences. More research is needed.

Epidemiology

- The estimated incidence is 1 in 3,000 births.[^2]
- Finland has a comparatively high figure of 1 in 2,500 births.
- The recurrence risk in subsequent pregnancies of oesophageal atresia/TOF that is not part of a syndrome of problems is <1%.[^3]

Associations

In more than 50% of babies, oesophageal atresia is present with other anomalies. Associated anomalies are more likely if there is isolated oesophageal atresia and, in such cases, can occur in up to 65%.[^3] They include:

- The VACTERL syndrome - the presence of three or more of:
  - Vertebral defects: including single or multiple hemivertebrae, scoliosis or rib deformities.
  - Anorectal malformations: including imperforate anus and cloacal deformities.
  - Cardiovascular defects: ventricular septal defects (most common), Fallot's tetralogy, patent ductus arteriosus, atrial septal defects, aortic coarctation, right-sided aortic arch, single umbilical artery, and others.
  - Tracheo-oesophageal defects.
  - (o)Esophageal atresia with or without TOF.
  - Renal abnormalities: including renal agenesis, horseshoe kidney, polycystic kidneys, urethral atresia and ureteral malformations.
  - Limb deformities: including radial dysplasia, absent radius, radial-ray deformities, syndactyly, polydactyly, lower-limb tibial deformities.

- The CHARGE association:
  - Coloboma
  - Heart defects
  - Atresia choanae
  - Retarded development
  - Genital hypoplasia
  - Ear abnormalities

- Chromosomal abnormalities:
  - Trisomy 13, 18 and 21.

- Other associations
  - DiGeorge's syndrome.
  - Neurological defects, including neural tube defects and hydrocephalus.
  - Gastrointestinal defects, including duodenal atresia, omphalocele and Meckel's diverticulum.
  - Pulmonary defects, including diaphragmatic hernia and pulmonary agenesis.
  - Genitalia defects, including hypospadias and undescended testes.

Of those with associated anomalies, 35% have cardiovascular defects, 20% genitalourinary defects and 20% gastrointestinal defects.

Presentation

Antenatally

- Diagnosis may be suspected antenatally because of polyhydramnios and an absent fetal stomach bubble detected on ultrasound.
- The prenatal detection rate using ultrasound if there are no other associated abnormalities was around 45% in one study.[^4] However, it does not allow for a definite diagnosis of oesophageal atresia/TOF.[^5]
- Associated ultrasound abnormalities may be present such as cardiac defects.
- The fetus is usually small for gestational age.
- Premature labour can occur.
- Karyotyping should be carried out if suspected because of the high association with trisomy 18.
Postnatally

- A baby with oesophageal atresia ± TOF classically presents with respiratory distress, choking, feeding difficulties and frothing in the first few hours after birth.
- Swallowing cannot occur due to the lack of patency of the oesophagus.
- Passing of a nasogastric tube is not possible.
- There is an overflow of saliva and aspiration can occur. If there is a TOF present, saliva ± gastric secretions can pass directly to the bronchial tree.
- H-type fistulae usually present later in infancy as there is no ‘blind end’ to the oesophagus and the child is able to feed. Children usually present with a recurrent cough on feeding or recurrent chest infections.

Oesophageal atresia ± TOF should be considered whenever a baby develops feeding and respiratory difficulties in the first few days of life.

Investigations

- CXR: this can show the heart size and shadow, any vertebral and rib abnormalities and can be used to assess the lung fields. The presence of air below the diaphragm should be assessed. If there is no air seen in the gastrointestinal tract, it is likely that there is isolated oesophageal atresia with no TOF. Air can also be injected to distend the upper oesophageal pouch prior to X-ray so that the blind ending pouch may be seen. If attempt has been made to pass a nasogastric tube, it can be seen curling up in the upper oesophageal pouch.
- Imaging of the renal tract: this is important to assess any problems of the urogenital tract.
- Echocardiography: can assess the heart.
- Limb X-rays: if limbs appear abnormal then X-ray is required.
- Ultrasound examination of the spine: can assess possible tethering of the spinal cord.
- A ‘gap-o-gram’ may be necessary to assess the distance between the proximal and distal parts of the oesophagus.

Management

- A multidisciplinary approach involving surgeons, physiotherapists, respiratory physicians, dieticians and speech therapists is best.
- If suspected antenatally, all babies with oesophageal atresia ± TOF should be delivered somewhere with ready access to a paediatric surgical unit.
- The basis of management is surgery to correct the anatomical abnormality.
- Surgery is carried out either immediately, as a delayed repair or as a staged repair depending on other factors such as birth weight and other associated conditions (principally cardiac abnormalities).
- It may be necessary to assess and manage other congenital anomalies as well.
- Until surgery, supportive treatment is needed to allow hydration/feeding and to prevent aspiration.
- A ‘replogle tube’ is passed through the nose into the proximal oesophageal pouch to provide drainage.

Oesophageal atresia with TOF

- Pre-operative bronchoscopy can be helpful in identifying and locating fistulae.
- An open thoracotomy is usually performed, the fistula is tied off and an oesophageal anastomosis is created between the disconnected upper and lower oesophageal segments.
- Sometimes, the gap between the segments can be long (so-called ‘long-gap’) and various procedures have been developed to deal with this. The Foker technique for long-gap oesophageal atresia has been approved by the National Institute for Health and Care Excellence (NICE). It involves applying traction sutures to the oesophageal ends to stimulate a degree of elongation each day and eventually allow primary anastomosis. Other procedures have been developed for long-gap oesophageal atresia including pulling the stomach partially up into the thorax, or using colon to join the oesophageal ends. However, the ‘native’ oesophagus approach is preferred.
- Minimally invasive surgical techniques have also been developed.

Isolated oesophageal atresia

- Immediate management involves a gastrostomy so that feeding is possible. Suctioning of the blind ending oesophagus is necessary to prevent aspiration and protect the airway. This should continue until surgery is performed. Prophylactic antibiotics may also be needed.
- Definitive treatment involves either creating an anastomosis between the native oesophageal segments (as described above) or using colon or stomach to enable the repair. ‘Long-gap’ procedures may be necessary.

H-type fistula

- Surgery is usually performed via the neck and the fistula is divided and repaired. There is a risk of recurrent laryngeal nerve injury.
- Laser repair has also been used.

Complications

After repair of oesophageal atresia in infancy, gastro-oesophageal reflux, oesophageal dysmotility and respiratory problems are common and significant oesophageal morbidity extends into adulthood.
Early complications include:

- Leakage of the anastomosis.
- Recurrent TOF.
- Anastomotic stricture (may require dilatation and a few may need resection of the stricture). [12]
- Feeding problems and poor weight gain.

Later complications include:

- Respiratory complications:
  - Tracheomalacia can lead to a 'TOF cough' (a harsh barking cough).
  - Recurrent chest infections can lead to bronchiectasis and irreversible lung damage.
  - Respiratory morbidity tends to improve as the child ages.
  - Management includes the use of antibiotics, physiotherapy and treatment of gastro-oesophageal reflux to minimise aspiration. Bronchodilators and inhaled steroids may be needed. [2]

- Gastro-oesophageal complications:
  - Gastro-oesophageal reflux is very common. This can contribute to stricture formation, respiratory problems and can lead to oesophagitis.
  - Management is using feed thickeners, H2 receptor antagonists, proton pump inhibitors and prokinetic drugs. [2] Investigation and anti-reflux surgery are needed in some.
  - Reflux symptoms usually improve with age.
  - The potential risk of Barrett's oesophagus with subsequent oesophageal carcinoma means that some suggest long-term monitoring. [13]
  - Oesophageal dysmotility can be seen on manometry. It can lead to problems with swallowing and choking.

- Other complications:
  - There may be other complications depending on any associated anomalies.

Prognosis

- This depends on associated anomalies and their complications.
- Overall survival now exceeds 90% in dedicated centres. [14]
- Associated congenital heart defects and low birth weight can affect survival.
- Catch-up growth normally occurs after successful treatment.
- Early mortality is usually due to cardiac and chromosomal abnormalities.
- Late mortality is usually due to respiratory complications.

Further reading & references

- TOFS (Tracheo-oesophageal Fistula Support)
  7. Foker technique for long gap oesophageal atresia; NICE Interventional Procedures Guidance, January 2006

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