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Best practice guidelines

Oesophageal atresia and tracheo-oesophageal fistula

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ABSTRACT

Oesophageal atresia with tracheo-oesophageal fistula is a relatively common congenital anomaly occurring in around 1:2500 births. The aetiology and embryology of the condition remain unclear, whilst associations with other significant anomalies are common. Studies in rodent models are contributing to our understanding of the condition. Advances in surgical care and neonatal management have improved survival considerably to around 90%. Long-gap and isolated oesophageal atresia present significant management challenges. Post-operative and long-term complications including oesophageal stricture, gastro-oesophageal reflux and respiratory compromise however remain relatively common and continue to pose a challenge for the ongoing management of patients.

Contents

1. Definition/incidence

Oesophageal atresia and tracheo-oesophageal fistula have an incidence of around 1:2500 live-births [1]. It may be divided anatomically into 5 types (see Fig. 1) with the most common being oesophageal atresia with a distal tracheo-oesophageal fistula — found in around 85% cases. The condition consists of a discontinuity or atresia of the oesophagus; with the majority of infants exhibiting a connection or fistula between the oesophagus and trachea. The exceptions to this are children born with an isolated oesophageal atresia and those with an H-type tracheo-oesophageal fistula. The latter of these tends to present after the neonatal period, is challenging to diagnose and is treated by a different operation. Thus, its management will not be discussed further here.

2. Embryology

During early development the airway develops as a diverticulum from the primitive foregut. Development starts as early as 4 weeks of gestation. Once the diverticulum is formed, it extends caudally into

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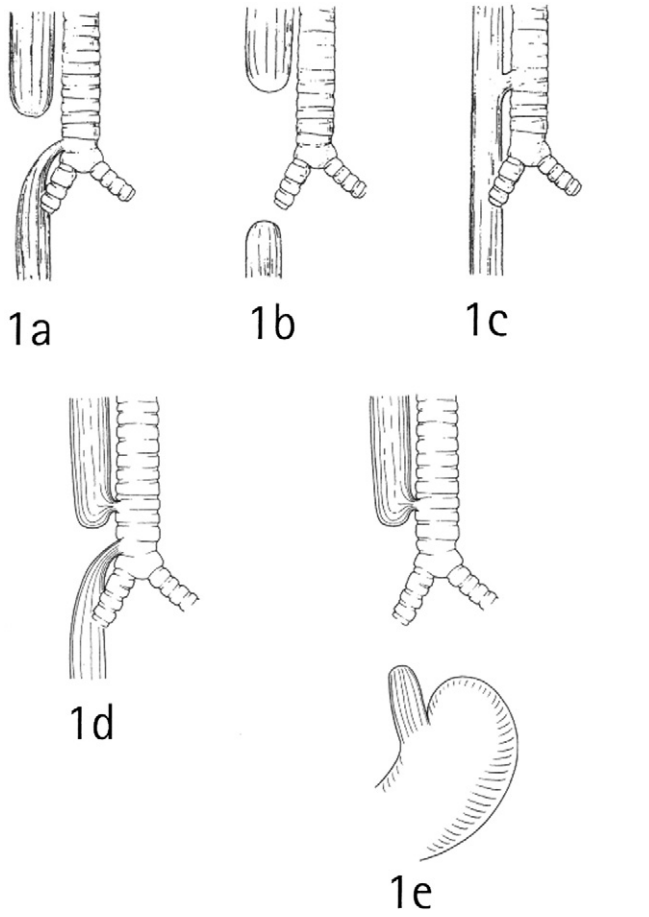

Fig. 1. Line drawing showing the five anatomical variants of congenital oesophageal atresia/tracheo-oesophageal fistula. 1a: oesophageal atresia with distal fistula (85% cases); 1b: oesophageal atresia without fistula (75); 1c: H-type TOF (4%); 1d: oesophageal atresia with proximal and distal fistula (3%); 1e: oesophageal atresia with proximal fistula (1%).

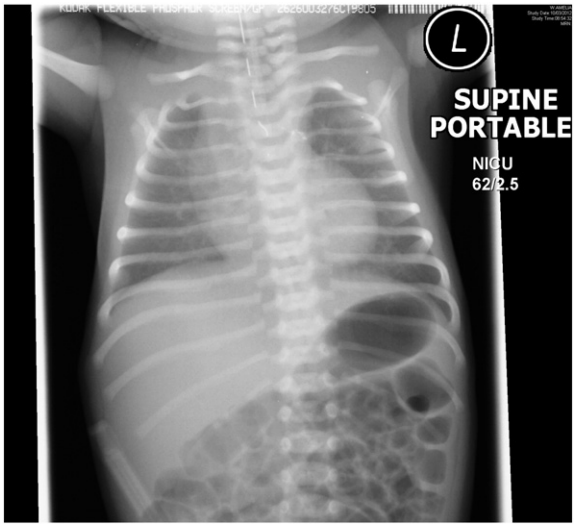

Fig. 2. Chest radiograph of an infant with OA-TOF. The nasogastric tube can be seen terminating in the upper pouch. Sub-diaphragmatic air indicates the presence of a distal tracheo-oesophageal fistula.

the splanchnic mesoderm and divides into two tubules which will form the right and left main bronchi. Simultaneously the formation of the trachea-oesophageal groove begins to separate the two tubes in a rostro-caudal direction. This common origin provides one explanation for the embryology of tracheo-oesophageal fistulae — presumed to result from a failure of invagination of the lateral tracheo-oesophageal grooves. The teratogenic Adriamycin rodent model of OA-TOF has been very useful in helping to uncover some of the developmental features of the condition. However the embryology remains unclear.

3. Diagnosis

In the vast majority of infants the diagnosis is made in the early postnatal period. Antenatally, polyhydramnios along with a small stomach has around a 55% predictive value for oesophageal atresia. In addition the presence of recognised associated anomalies (see below) may raise the possibility of the diagnosis, however it is not possible to definitively diagnose OA-TOF on an antenatal ultrasound scan.

Diagnosis is therefore made when attempts are made to feed the infant. The baby may then be described as 'bubbly', be unable to tolerate feeds, may experience choking episodes or become cyanotic when attempting to feed. If the diagnosis is suspected, an NG tube should be passed. Failure to pass a tube into the stomach, along with a characteristic x-ray (Fig. 2) is diagnostic of OA-TOF. Caution should however be exercised to ensure the NG tube is of sufficient calibre to not simply coil in the back of the pharynx, and that it is pushed down as far as possible to allow the diagnosis to be made — without of course damaging the upper pouch. Surgical teaching is for a 10Fr nasogastric tube to be passed — which typically cannot be advanced beyond 10 cm. The initial film will allow assessment of the presence or absence of distal gas, which indicates whether a distal fistula is present. The absence of gas in sub-diaphragmatic bowel loops indicates that a distal fistula is not present — this raises concerns of a long oesophageal gap; and the possibility of technical difficulties at surgery. If there is ongoing doubt regarding the diagnosis, on rare occasions a contrast study may be helpful. However, extreme care should be taken as there is a high risk of aspiration of contrast during the study.

4. Associated anomalies & genetic associations

OA-TOF may be associated with other anomalies, and these should be sought during the early assessment. Most common is the VACTERL association. This mnemonic indicates defects in Vertebrae, Anorectal malformations, Cardiac anomalies; Tracheo-Esophageal fistula; Renal abnormalities and Limb defects (most commonly radial hypoplasia or agenesis). Many other associations have been noted with varying degrees of frequency, including duodenal atresia, intestinal malrotation, and other skeletal anomalies. Other associations include the CHARGE syndrome and trisomy 18. To date no definitive genetic basis for OA-TOF in humans has been identified. Around 1% of cases show familial inheritance; the remainder are sporadic diagnoses [2]. Murine models including the Shh−/− knockout demonstrate a phenotype including OA-TOF and have been used to study some aspects of the condition. However, patients with abnormalities in this pathway, including mutations in the GLI family of transcription factors (downstream effectors of the SHH pathway) do not seem to exhibit OA-TOF as part of their phenotype. More recently microdeletions of the FOX gene cluster at 16q24.1 have been found in patients with wide-ranging abnormalities including respiratory and foregut malformations [2]. Further detailed investigations are needed to elucidate the exact nature of any genetic abnormality underlying OA-TOF in isolation.

A full clinical examination including inspection of the perineum, spine and assessment of the limbs is essential. Babies diagnosed with OA-TOF should also undergo echocardiography prior to anaesthesia, with ultrasound of the renal system and spine being performed in the early neonatal period.