CASE REPORT

Tracheal agenesis in a new born: lessons learnt

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SUMMARY

Tracheal agenesis is a rare but fatal congenital tracheal malformation. Lack of prenatal symptom and a typical clinical presentation lead to failure to arrive at a correct diagnosis and confusion during resuscitation. We report a case of a newborn male child with type 2 tracheal agenesis. Despite a typical presentation, diagnosis was delayed after unsuccessful intubation, examination under anaesthesia and emergency tracheostomy. The embryology, diagnostic criteria and potential treatment options are discussed. This case report is valuable in increasing awareness of this rare condition and will help us in being better prepared in managing these children. Future studies should aim to find the optimal replacement for the tracheal.

BACKGROUND

Tracheal agenesis (TA) is a rare potentially lethal congenital anomaly with an incidence of 1 per 50 000 newborns.¹ It is characterised by complete or partial absence of trachea below the larynx with or without tracheoesophageal fistula (TOF). So far close to 200 cases have been reported in literature with only a handful of cases reporting long-term survival.² This case report adds to and extends the data available and also highlights the challenges encountered in diagnosis and management of this condition.

CASE PRESENTATION

A newborn male baby of 1500g was born at 34 weeks of gestation to a primigravida by caesarean section. The pregnancy was complicated by polyhydramnios but routine antenatal ultrasound did not pick up any abnormality. The baby had immediate respiratory distress but despite multiple attempts at intubation he could not be intubated in spite of good visualisation of the glottis. Resuscitation was done with bag and mask ventilation and an urgent otolaryngology consult was sought.

INVESTIGATIONS

Endoscopic assessment in the neonatal intensive care unit (ICU) revealed complete atresia of the larynx from the subglottic level. Subsequently, the child was successfully ventilated by an endotracheal tube in the oesophagus, suggesting the presence of a TOF. The child was transferred to the operating table for a detailed assessment. Intraoperatively an oesophagoscopy was done to look for a TOF; however, none could be identified on table despite ventilation occurring through the oesophagus.

DIFFERENTIAL DIAGNOSIS

The child was found have other anomalies such as imperforate anus, atrial septal defect, patent ductus arteriosus and a unilateral dysplastic kidney suggestive of a possible VACTERL anomaly.

TREATMENT

During examination he had recurrent episodes of desaturation, tracheostomy was attempted but no trachea could be identified, confirming the diagnosis of TA. Repeat oesophagoscopy confirmed the presence of tracheostomy tube in the oesophagus.

OUTCOME AND FOLLOW-UP

Despite multiple attempts at resuscitation, the child died on table. Postmortem examination revealed a complete absence of cervical and thoracic trachea with a small pinhole opening in the anterior wall of the oesophagus giving origin to both the bronchi suggestive of a type 2 TA (figures 1 and 2). The child died probably due to mucus plugging of this small pinhole opening which could not be identified on oesophagoscopy but only on postmortem analysis.

DISCUSSION

TA is a rare congenital anomaly, first described in the literature by Payne. Floyd *et al* classified TA based on the degree of development and type of fistulous connection with the oesophagus into three types (figure 3). In type 1, the stump of the distal trachea attaches to the anterior oesophageal wall, in type 2 it is the carina with the two bronchus which attaches while in type 3 there are two separate attachments for the two bronchi.^{3,4}

TA is best explained by the recent theory that states that the lower respiratory tract develops as a respiratory diverticulum from the ventral aspect of the foregut which then elongates caudally to form the trachea. This is different from the earlier accepted theory that caudocranial fusion of the lateral ridges results in division of the foregut into ventral trachea and dorsal oesophagus. The new hypothesis is based on analysis of anomalous TE development in doxorubicin-exposed rat embryos. TA occurs with the development of a ventral rather than a dorsal pouch from the upper foregut, resulting in an atretic proximal trachea, while dorsal to that the foregut differentiates into oesophagus.^{5,6} Arrested elongation of the trachea results in type 1 while arrested elongation with fusion to form carina results in type 2 and without fusion to type 3.⁷ The exact aetiology is not known but genetic factors have been implicated.¹

Prenatal diagnosis is difficult unless TOF is absent, in which case the fetus will have features of

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Figure 1 Postmortem view of the thorax showing complete absence of trachea and a blindly ending larynx (yellow arrow).

congenital high airway obstruction syndrome (CHAOS). CHAOS is suspected when there is polyhydramnios and is characterised by enlarged hyperechogenic lungs, flattened diaphragms and fluid-filled dilated trachea and bronchi. In the presence of a TOF, these signs will not be present, making prenatal diagnosis difficult. Presence of polyhydramnios and other congenital anomalies may be the only clue for the clinician. In such cases, prenatal MRI may provide a definitive diagnosis.⁸

Postnatal diagnosis of this condition is usually a respiratory emergency. It is considered usually in a preterm low birthweight baby with respiratory distress with no audible cry and inability to intubate despite good visualisation of the glottis.⁹

TA may present as a single-organ malformation or may be a part of other congenital abnormalities association. Most cases are associated with either the 'VACTERL' association (vertebral defects, anal atresia, TOF with oesophageal atresia and radial or renal dysplasia, plus cardiovascular and limb defects) 'VATER' association (without cardiovascular association) or the 'TARCD' association (TA or atresia, radial ray defects, complex congenital cardiac abnormalities and duodenal atresia).¹⁷

When prenatal diagnosis is suspected, a planned EXIT procedure can be done for an airway control during the initial assessment.¹⁰ For immediate resuscitation, oesophageal intubation is a temporary solution. In Floyd's type I agenesis, a small endotracheal tube placed directly in the distal tracheal stump under bronchoscopic guidance is the best bet. In Floyd's type II and III agenesis, oesophageal intubation can maintain the infant's airway temporarily; however, the precarious position of the tube and the escape of gases into the stomach can lead to abdominal distension further worsening the respiratory distress. The additional surgeries needed for palliation may include distal

Figure 2 View from posteriorly of the anterior wall of the oesophagus showing a probe in the small pinhole opening (white arrow) leading into both lungs and the lower probe (yellow arrow) leading into stomach. Blue star points to lungs.

oesophageal banding, transection at the cervical oesophagus with oesophagostomy and gastrostomy for feeding. Postnatal diagnosis as mentioned is usually a respiratory emergency. Securing the airway if possible and resuscitation are the immediate priorities. Once those are achieved, CT scan is ideal in the diagnosis of TA both for diagnosis and further treatment planning. Airway ventilation manoeuvres to improve the aeration of the collapsed airways in TA facilitate better visualisation.¹¹

Outcomes in terms of definitive surgical repair for this condition have been abysmal.

Mortality of these children has been reported to be around 85% with a few isolated cases of short-term survival as summarised by Mohammed *et al*.² In a type 1 TA, Barorcini *et al* in 2004, Watanabe *et al* in 2008 and Usui *et al* in 2010 have reported survival (10 months, 28 months and 3 years follow-up, respectively).^{12–14} Park *et al* in 2015 reported survival up to almost 1 year for a type 2 TA while in the same year Tazuke *et al* reported survival ranging from 77 to 109 months for four children (two type 1, one type 2, one type 3).^{15 16} Recently, Densmore *et al* reported survival (follow-up 146 days) in type 2 TA with neonatal trachealisation and oesophago-carinoplasty.¹⁷ Overall neo trachealisation of the oesophagus has been the surgery of choice; however, the outcomes still remain dismal for this rare condition. These infants have a stormy postoperative course including multiple surgeries and prolonged postoperative course. An experienced anaesthetist familiar with the lesion, a dedicated surgical and ICU team and proper communication among all are essential in the management of these complex cases.¹⁸

Figure 3 Classification scheme of tracheal agenesis.

The oesophagus is a soft collapsible tube and tends to collapse with negative inspiratory effort. Internal or external splints to stiffen the oesophagus have been tried but they have their own set of problems.¹⁵ Internal or indwelling stents have the problem of granulations and infection and need to be cleaned periodically in a controlled setting. Accidental extubation can have lethal consequences. External stents, on the other hand, can cause problems such as erosion into nearby structures as well as airway perforation.

The ideal repair would be possible if a homologous or synthetic tracheal prosthesis were available for replacement. The oesophagus is not the ideal replacement for an absent trachea as it lacks the rigidity of the tracheal rings as well as the normal ciliated epithelium necessary for tracheal functions. Internal or external supports can give rigidity to the oesophagus but not the appropriate respiratory epithelium. They also may not be conducive to accommodate for the growth of the child.

At present, a curative treatment of TA is not available. Options in the future include tissue engineering offering a hope towards tracheal transplantation. Translational research and animal experiments may offer directions to a possibility of an ideal tracheal replacement with required rigidity, humidification, ciliary function and options of growth with the child.¹⁷ Till then TA remains a potentially lethal condition which can be a major burden to any healthcare system. This is more so in resource-poor countries like ours where healthcare is not state financed but financed by the patient himself. A dedicated team of multiple specialties is needed in addition.

This case report highlights that a potentially lethal anomaly like TA can exist in a presumably uncomplicated pregnancy. The

presence of respiratory distress with inaudible cry and inability to intubate should raise suspicion of this condition. Despite a detailed oesophagoscopy in the operating table, sometimes the opening is so small, like in our case, that it may be missed. Proper counselling of the parents is needed immediately after birth to prepare the parents for this potentially lethal condition.

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Learning points

- Prenatal diagnosis of this rare lethal condition is difficult in the presence of a tracheoesophageal fistula.
- The presence of respiratory distress in a low birthweight newborn with inaudible cry and inability to intubate despite good visualisation should alert the clinician to this condition.
- Oesophageal intubation offers a temporary solution for immediate resuscitation.
- Definitive surgical repair is challenging, expensive with prolonged and multiple hospitalisations and with poor outcomes.
- Options in resource-poor setting are limited.
- Proper counselling of the parents is needed.

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