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Case Report

Paediatrics Section

Floyd Type II Congenital Tracheal Agenesis in a Preterm Neonate: A Rare Case Report

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ABSTRACT

Tracheal agenesis is a rare and life-threatening airway malformation, and currently, there is no curative treatment available. The described case involves a preterm male newborn at 30 weeks of gestational age who could not be intubated during resuscitation, despite multiple attempts. However, the baby could be ventilated after oesophageal intubation. Tracheal stenosis/ atresia was suspected, and airway evaluation was performed using a thin, flexible fiberoptic bronchoscope. No tracheal opening could be identified, and upon introducing the bronchoscope into the oesophagus, a triluminal opening was found, through which the scope could not be further navigated. To further delineate the anatomy, a Contrast-enhanced Computed Tomography (CECT) thorax was performed, revealing the absence of a tracheal lumen and the communication of both bronchi at the carina with the oesophagus. Unfortunately, the baby succumbed to the illness after three days. Tracheal agenesis is an anatomical malformation that typically presents as respiratory distress and the absence of an audible cry at birth. Attempts to establish a definite airway are unsuccessful, which may result in early neonatal death. Oesophageal intubation may temporarily establish ventilation until palliative surgery is performed.

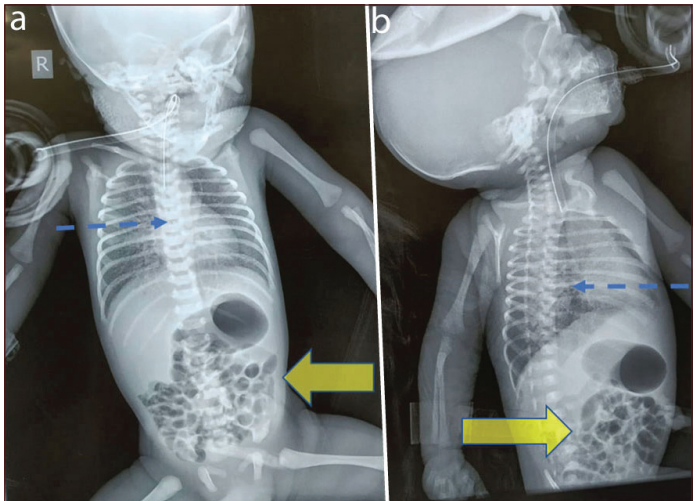
Keywords: Airway malformation, Polyhydramnios, Tracheo-oesophageal fistula

CASE REPORT

A male preterm baby, born at 30 weeks of gestational age with a birth weight of 1.65 kg, exhibited inadequate respiratory effort at birth, necessitating immediate resuscitation. Despite attempts at endotracheal intubation during neonatal resuscitation, the procedure proved unsuccessful. Consequently, the baby was admitted to the Department of Neonatology for specialised care and further management. He was delivered to a 20-year-old primigravida woman. An antenatal ultrasound was conducted at 27 weeks of gestation, which suggested polyhydramnios {Amniotic fluid index (AFI)- 32 cm}. The mother was anaemic with a haemoglobin of 6.07 gm/dL; therefore, received two units of packed Red Blood cells (RBC) transfusion a month before delivery. She developed foetal distress at 30 weeks of gestation and went into premature labour.

The baby was delivered after an emergency caesarean section. The baby did not cry immediately after birth and was cyanosed, with Appearance, Pulse, Grimace, Activity and Respiration (APGAR) scores of 1/10, 5/10 and 7/10 at 1, 5 and 10 minutes, respectively. After initial steps of resuscitation including warmth, drying, stimulation, positioning and suction, the baby's heart rate was 60 beats per minute (bpm), and gasping respiration was present. Bag and mask ventilation was initiated, and after 30 seconds, the baby's heart rate increased to 80 bpm. Bag and mask ventilation was continued for two minutes, but the heart rate remained below 100 bpm, and Saturation of Peripheral Oxygen (SpO_2) remained between 40% and 50%. An attempt was made to perform endotracheal intubation using a 2.5 mm endotracheal tube. During laryngoscopy for intubation, the vocal cords were visible normally, but there was no passage past the vocal cords. During intubation attempts, accidental oesophageal intubation occurred, which surprisingly led to bilaterally symmetrical chest rise along with gastric distension with each breath. The saturation improved to 90-95% within minutes, and the baby was shifted to the Neonatal Intensive Care Unit (NICU) with oesophageal intubation.

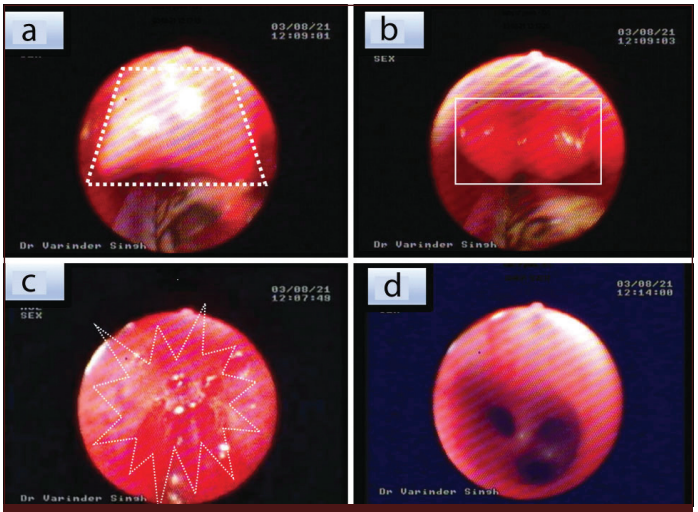
In the NICU, the baby was maintained with oesophageal intubation on Synchronised Intermittent Mandatory Ventilation (SIMV) mode, with Positive End Expiratory Pressure (PEEP)-6 cmH_2O, Peak Inspiratory Pressure (PIP)-20 cmH_2O, Fraction of Inspired Oxygen (FiO_2)-40%, respiratory rate-50 cycles per minute, and Inspiratory Time (tI)-0.34 seconds, and SpO_2 was maintained between 90-95%. However, frequent gastric decompressions were required due to gaseous distension. Difficulty in passing an orogastric tube was also noticed during some attempts, whereas sometimes it was easily placed. A chest skiagram confirmed the orogastric tube's position in the stomach. A possibility of tracheal atresia/ stenosis with Tracheo-oesophageal Fistula (TOF) was considered. The chest X-ray Anteroposterior (AP) view [Table/Fig-1a] showed normally expanded bilateral lung fields with the carina at T6. The lateral view [Table/Fig-1b] suggested the endotracheal tube in the



[Table/Fig-1]: Babygram (a) Anteroposterior revealed normal aeration of bilateral lungs with carina at T6 (dashed arrow) and normal abdominal bowel gas pattern (yellow arrow); (b) Lateral view revealed endotracheal tube in oesophagus and no tracheal shadow in thorax.

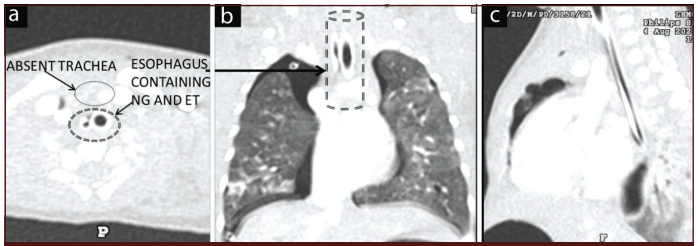
oesophagus and no tracheal shadow in the thorax. At 12 hours of life, the baby developed fluid-responsive shock.

The first-line antibiotics (Piperacillin-tazobactam 100 mg/kg/dose q 12 hours and amikacin 12 mg/kg/dose q 36 hrs) were initiated considering probable sepsis. Venous blood gas analysis showed worsening hypercapnia, respiratory and metabolic acidosis with a pH of 6.93, Partial Pressure of Carbondioxide (pCO_2) of 73, and Bicarbonate (HCO_3) of 11. A bronchoscope (3.6 mm) was used for airway visualisation. The epiglottis and arytenoids were normally formed; however, no opening was visualised through the laryngeal inlet. As the child could be ventilated through the oesophagus, an evaluation of the oesophagus was conducted using the bronchoscope. At approximately 14 cm from the mouth, the oesophagus divided into three openings, one of which showed constant bubbling of air. None of the openings were large enough to allow the 3.6 mm bronchoscope to advance further [Table/Fig-2a-d].

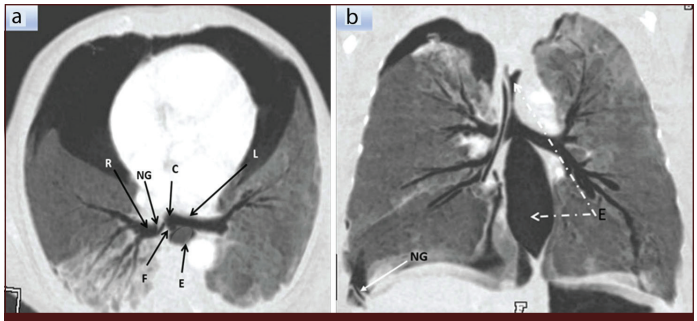


[Table/Fig-2]: Bronchoscopy/ upper gastrointestinal endoscopy a,b) normally formed epiglottis and arytenoids; c) The larynx showed a complete web with no opening seen beyond; d) The lower end of the oesophagus showed three openings, of which one of the opening continued into the stomach.

A CECT thorax was planned to further detail the anatomy. It showed non visualisation of the tracheal lumen in the superior mediastinum with the presence of a common bronchus (carina) connecting the left and right main bronchi to the oesophagus. There were bilateral pneumothoraces with the orogastric tube seen coiled in the right pleural space. The orogastric tube was observed coursing through the right main bronchus and the posterior basal segment branch of the right lower lobe bronchus, piercing through the pleura with its extension into the right pleural cavity [Table/Fig-3a-c,4a,b]. Although the initial placement of the orogastric tube was successful, as confirmed in the chest skiagram, probably during one of the repeat attempts for the orogastric tube placement, it passed through the right bronchial opening and pierced the pleura, leading to pneumothorax.



[Table/Fig-3]: CECT lung window axial, coronal, sagittal reformat (a-c): Trachea is absent in the superior mediastinum. Both Endotracheal Tube (ET) and Nasogastric Tube (NG) are seen within the proximal dilated oesophagus.



[Table/Fig-4]: Minimum intensity projection (a,b): The Right (R) and Left (L) bronchus are seen arising from a common bronchus (carina) with presence of a carino-oesophageal Fistula (F). The NG is seen to enter into the right main bronchus after passing through the fistula between the Carina (C) and the oesophagus (E) and is piercing the pleura with its extension into the right pleural cavity. Bilateral pneumothorax is seen and the distal oesophagus appears dilated.

Based on the above findings, a diagnosis of Floyd's type II tracheal agenesis with tracheo-oesophageal fistula was made, and the parents were explained the prognosis of the child. The differential diagnoses considered were laryngeal atresia, severe glottis web, congenital tracheal stenosis and Vertebral, Anal, Cardiac, Tracheoesophageal fistula with or without oesophageal atresia, Renal and Limb defects (VACTERL) syndrome, which were ruled out after delineating the anatomy with the help of bronchoscopy and chest CT scan. An ultrasound of the abdomen was performed to look for other associated anomalies. Bilateral hydronephrosis {Anteroposterior Diameter (APD) 10 mm on the left side and 20 mm on the right side} was observed, the urinary bladder was distended, and showed mobile internal echoes possibly due to cystitis. No other abnormality was detected on the ultrasound of the abdomen.

Bilateral intercostal drainage tubes were placed for the pneumothoraces detected in the CECT. At 38 hours of life, the baby developed fluid-refractory shock presented with cold extremities, tachycardia and a mean arterial pressure of 24 mmHg, for which a dopamine infusion was started at 10 µg/kg/min. Sequentially, injection adrenaline, noradrenaline and vasopressin were also gradually added in order to maintain the mean arterial pressure. Antibiotics were upgraded to meropenem 20 mg/kg/dose 12 hourly and amikacin 12 mg/kg/dose 36 hourly in view of clinical deterioration after sending blood culture, which was reported as sterile At 48 hours of life, the baby also developed sclerema neonatorum, pCO_2 retention kept on worsening despite optimising ventilator settings. An increase in pCO_2 (123 mmHg in venous blood gas) and worsening shock led to severe acidosis (pH=6.87). The clinical condition further deteriorated, and the baby had a cardiac arrest and died at 70 hours of life. Whole exome sequencing was advised but refused by the parents due to financial constraints.

DISCUSSION

Tracheal agenesis is a rare and fatal congenital anomaly first described by Payne WA in 1900 [1]. The prevalence is less than 1 in 50,000, with a male to female ratio of 2:1 [2]. It usually goes undetected in the prenatal period unless associated with the absence of a TOF. If there is no fistula, the foetus presents with Congenital High Airway Obstruction Syndrome (CHAOS). The presence of a fistula allows the lung fluid to pass through the intestines, and therefore prevents the manifestation of CHAOS, as seen in the present case. Most cases, like the present case, are associated with antenatal polyhydramnios, which raises the

suspicion of a tracheal malformation [3]. However, the diagnosis can be established by a detailed ultrasonogram or more certainly by foetal Magnetic Resonance Imaging (MRI).

Floyd's classification of Tracheal Agenesis (TA) is the most commonly used system [4]. It divides the condition into three main types: Type-I, short distal tracheal remnant with TOF. Type-II, no tracheal remnant; bronchi communicate at the carina with TOF. Type-III, bronchi communicate through separate fistulas with the distal oesophagus.

The exact pathogenesis of tracheal agenesis is unknown. Several theories suggest an insult during the early embryologic differentiation of the respiratory tract [5]. Different theories have been suggested to explain the cause of tracheo-oesophageal anomalies. In addition to environmental factors, various genetic abnormalities in animal models, including the (conditional) inactivation of Gli2, Gli3, Shh, Foxf1, and beta-catenin genes, manifest aberrations such as tracheal agenesis or abnormal septation of the foregut. The involvement of Bone Morphogenetic Protein (BMP) type-I receptor genes has been proposed as a significant factor in similar observations in mouse models [6]. However, no causal gene has been identified in human TA patients yet.

The clinical presentation is immediate after birth with respiratory distress, inaudible cry and failure of intubation. It is also associated with preterm birth and poor APGAR scores. Attempts to establish an airway by tracheostomy fail. In cases of TA accompanied by a TOF, bag and mask ventilation and oesophageal intubation may temporarily improve the respiratory status, but it will also lead to simultaneous abdominal distension, causing frequent deteriorations. Mechanical ventilation is usually required to sustain life. To date, approximately 200 cases of tracheal agenesis and more than 100 cases of type 2 tracheal agenesis have been reported worldwide, and only two babies have been reported to survive without mechanical ventilation after airway reconstruction surgeries [7,8]. There is also a high incidence of associated anomalies, which has led researchers to think that this is a part of VACTERL or Tracheal Agenesis or Laryngotracheal Atresia, Complex Congenital Cardiac Anomalies, Radial Ray Defects and Duodenal Atresia (TACRD) association [9].

Imaging can provide clues to the diagnosis, as a chest radiograph may show the absence of a tracheal shadow and a posteriorly placed oesophageal tube. CT scan with post-processing and multiplanar reconstruction can help in identifying the anatomy [10]. Endoscopy can show the absence of the trachea and a fistula between the oesophagus and the airway [11]. Often, a combination of techniques may be needed to know the extent and type of airway defect. The diagnosis can reliably be detailed only by operative or autopsy findings.

Tracheal agenesis is nearly always fatal in the neonatal period. Few case reports describe survival beyond infancy after palliative surgeries, with the longest reported survival being six years [12]. In prenatally diagnosed cases, an ex-utero intrapartum treatment can be planned, as suggested by Vaikunth SS et al., Oesophageal intubation can be used as a lifesaving and temporary measure to establish ventilation until surgical corrections are attempted [13]. Surgeries needed for palliation may include distal oesophageal banding, transection at the cervical oesophagus with oesophagostomy and gastrostomy for feeding. Usui N et al., performed a three-stage reconstruction for the airway and alimentary tract after the baby had a distal oesophageal banding with feeding gastrostomy at six hours of birth [7]. They created a proximal cervical oesophagostomy for salivary secretions and a distal cervical oesophagostomy for the airway connected to a ventilator through a tracheostomy cannula. At eight months of age, anastomosis of the upper mid-oesophagus and carinal trachea was done, followed by the reconstruction of the oesophagus by joining the cervical oesophagus and the lower oesophagus. Despite progress in surgical management, only two children were reported alive with a normal neurodevelopment after surgery [7,8].

Currently, there is no existing curative remedy for tracheal agenesis. Future exploration into translational research and animal studies might provide insights into the potential development of an optimal tracheal replacement, encompassing necessary attributes such as rigidity, humidification, ciliary function and adaptability to a child's growth [14].

CONCLUSION(S)

Tracheal agenesis is a lethal respiratory tract malformation with no definitive surgical correction available to date. In these cases, the conventional methods to establish the airway taught in Neonatal Resuscitation Program (NRP) (e.g., intubation, laryngeal mask airway, or tracheostomy) do not succeed. Having an insight into the condition is important, as oesophageal intubation can provide a temporary airway until palliative surgery is planned.

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