Case Report

DOI: 10.7860/IJNMR/2025/76919.2450

Paediatric Surgery Section

Tracheo-oesophageal Fistula with Partial Pulmonary Agenesis, Broncho-oesophageal Fistula, and Gastric Duplication in a Neonate: A Case Report on the Spectrum of Aberrations in Foregut Embryogenesis

NEEL AGGERWAL[1], UMESH B SINGH[2], SHEETAL UPRETI[3]

ABSTRACT

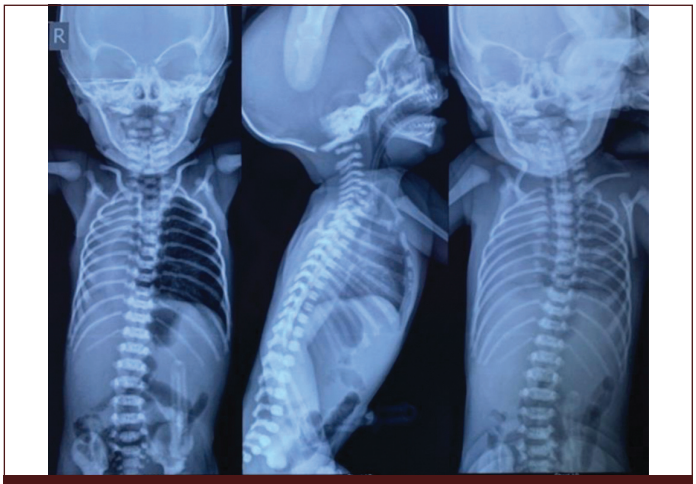
An erroneous connection between the trachea and oesophagus is the hallmark of Tracheo-Oesophageal Fistula (TEF), a congenital disorder that can provide serious complications for newborn care. TEF results from a defect in foregut embryogenesis that interferes with the trachea and oesophagus's natural separation during foetal development. Since, it frequently co-exists with other congenital conditions such as anorectal malformations, cardiac problems or skeletal abnormalities, early identification and interdisciplinary care are essential for the best results. A 36-week-old preterm low birth weight male neonate presented within 12 hours of birth, having respiratory distress, diagnosed as TEF, detected to have a rare combination of multiple congenital anomalies related to foregut development like partial pulmonary agenesis, bronchoesophageal fistula and gastric duplication in a preterm male neonate. The complexity of this presentation required coordinated surgical interventions including thoracotomy, oesophagostomy and excision of a gastric duplication cyst. To the greatest extent of the information, this was the first human case of its sort to be documented in the literature, exhibiting three foregut development abnormalities: gastric duplication, partial pulmonary agenesis and bronchooesophageal fistula. In the present case, the gastric duplication cyst was surgically removed during the TEF repair, highlighting the need of treating all abnormalities thoroughly to get better results. This case highlights the challenges and management strategies in addressing multiple anomalies in neonates with TEF.

Keywords: Congenital anomalies, Foregut embryological anomalies, Intestinal duplication, Oesophageal atresia

CASE REPORT

A 36-week-old preterm male neonate weighing 1.8 kg, presented to the paediatric emergency within 12 hours of birth with respiratory distress, excessive drooling of saliva, unable to feed with spells of chocking during trials of feed. There was no family history or any congenital anomaly reported in siblings. The neonate was referred from community health centre with chief complains of respiratory distress and excessive drooling of saliva. The case was born via normal vaginal delivery in a peripheral hospital. The foetus was antenatally supervised at some remote centre, only once at 26 weeks of gestation and ultrasonography detected a hypovolemic right hemithorax with mediastinal shift and a gastric bubble seen with polyhydramnios.

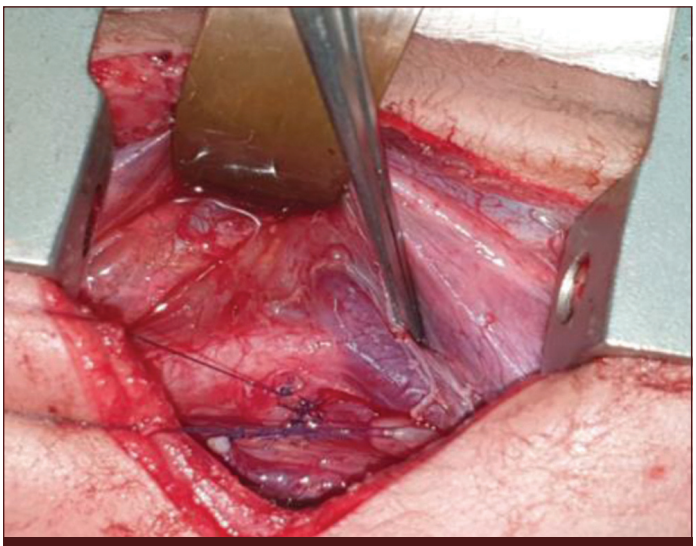
On examination, air entry on bilateral chest was normal with minimal crepitation, cardiac auscultation was within normal limits, nasogastric tube was coiling into neck indicating oesophageal Atresia (EA), anal opening was normal with meconium staining and no other external visible congenital abnormality was found. Systemic examination revealed normal s1 s2 on auscultation with slight shifting towards right and air entry present on bilateral chest. Preoperative haemoglobin was 13.4 mg/dL, Total Leukocyte Count (TLC) 6500, Na+ 135, K+ 4.3, chest X-ray confirmed coiling of nasogastric tube in neck and bowel gas pattern was normal confirming diagnosis of TEF and prepared for elective thoracotomy [Table/Fig-1].



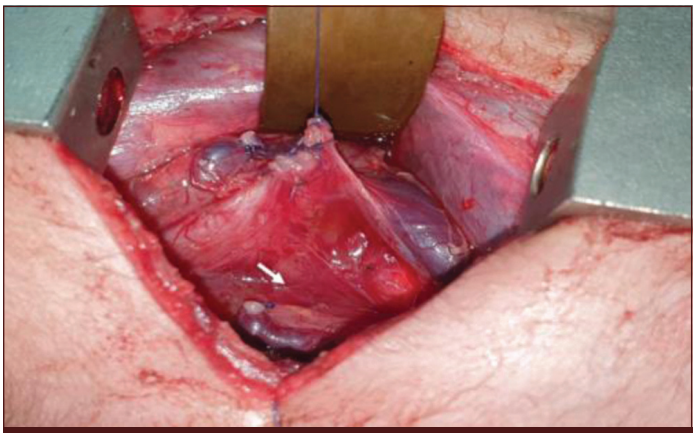
[Table/Fig-1]: Chest x-ray showing coiling of nasogastric tube in neck and normal bowel gas pattern.

On the operation table, exploration by incision on right chest wall using right posterolateral thoracotomy form 5th intercostal space, partial agenesis of the right upper and middle lung lobes was observed [Table/Fig-2]. The upper oesophageal pouch was blind ending about 3 cm above carina and lower oesophageal pouch communicated with the right lower bronchus instead of the trachea, indicative of EA and bronchoesophageal fistula [Table/Fig-3]. Due to the failure of oesophageal pouch approximation, the patient underwent cervical and abdominal oesophagostomy. The cervical

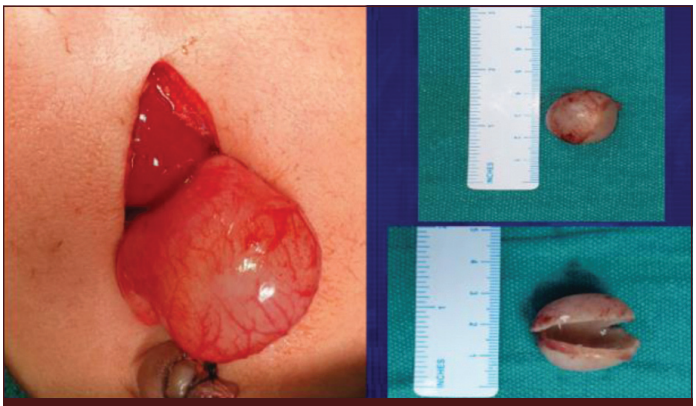
oesophagostomy created on right side of neck exteriorising upper oesophageal end about 1 cm above clavicle just medial to sternocleidomastoid. During abdominal exploration, a non communicating cystic duplication of 2×3 cm size along the greater curvature of the stomach was identified and excised [Table/Fig-4] along with completion of abdominal oesophagostomy of lower oesophageal end.



[Table/Fig-2]: Incision on right chest wall using right posterolateral thoracotomy form 5th intercostal space showing partial agenesis of the right upper and middle lung lobes.



[Table/Fig-3]: Image showing oesophageal atresia and bronchoesophageal fistula.



[Table/Fig-4]: Excision of non communicating cystic duplication of 2×3 cm size along the greater curvature of the stomach.

The neonate was shifted on ventilator in postoperative ward and subsequently to surgical neonatal intensive care unit. On postoperative day 3, blood culture revealed growth of *Klebsiella pneumoniae* and platelet counts fell to 25000/µL platelets. Intravenous broad spectrum antibiotics (inj. Meropenem 20 mg per kg every eight hourly and inj. Vancomycin 15 mg per kg every 12 hourly) started according to blood culture and sensitivity report along with platelet transfusion. However, patient condition continued deteriorating and pressures on ventilator continued to increase with worsening of lungs shown in skiagram. On postoperative day 7, case developed Disseminated Intravascular Coagulation (DIC), multiple site mucosal bleeding and ventilator parameters went high. Despite all efforts, the baby succumbed on postoperative day 8.

DISCUSSION

The TEF is a congenital anomaly characterised by an abnormal connection between the trachea and oesophagus. It commonly co-exists with other developmental abnormalities, including pulmonary agenesis and foregut duplications [1]. This case underscores the rarity and management complexities associated with such multiple anomalies. The index case presents a rare case of TEF in a neonate who presented within 12 hours of delivery with unique combination of several congenital defects pertaining to foregut development, including partial pulmonary agenesis, bronchoesophageal fistula and gastric duplication.

Unilateral pulmonary agenesis is exceedingly rare, with an estimated incidence of 1 in 15,000 autopsies in literature [2,3]. Association of pulmonary agenesis with TEF is reported in only 37 cases till date [4]. Its association with TEF has been reported in a limited number of cases, underscoring the diagnostic and management challenges posed by such complex anatomical anomalies. The prognosis for isolated pulmonary agenesis is generally poor and survival rates are further reduced in cases complicated by TEF [5]. Parelkar SV et al., demonstrated lethality of association of lung agenesis with EA and TEF in two cases managed surgically [6]. Literature reports only 16 surviving cases of TEF associated with pulmonary agenesis, highlighting the severity of this condition [7].

Only 17 cases has been reported till date in literature of TEF associated with foregut duplication cyst out of which only three cases underwent removal of cyst at the time of TEF repair [8]. Successful outcomes rely on prompt diagnosis, meticulous surgical intervention and multidisciplinary care to manage respiratory and nutritional challenges in these fragile neonates. Halilbasic A et al., reported a unique case with finding of unilateral pulmonary agenesis associated with large gastric duplication cyst and ventricular septal defect [9]. The closure yet common differential diagnosis of respiratory distress and difficult feeding in newborn may be prematurity of lungs in preterm newborns and congenital diaphragmatic hernia.

The co-existence of a foregut duplication cyst and pulmonary agenesis adds another layer of complexity to this case of TEF indicating anomalous foregut development. Till date, no case of TEF has been reported in literature having simultaneous anomalies of pulmonary agenesis and gastric duplication. However, animal models have given evidence of a common pathogenesis for foregut duplications and EA with TEF [10]. The accidental finding of foregut duplications in foetal rats with EA-TEF induced by Adriamycin provided an animal model indicating a possible relationship between these two entities pertinent to common embryogenesis from embryo foregut [10].

To the best of knowledge, this was the first case in human being reported in literature of its kind demonstrating three anomalies of foregut development including bronchoesophageal fistula, partial pulmonary agenesis and gastric duplication. Due to scarcity of reported cases in the literature, there is limited consensus on optimal management strategies of such cases. Surgical excision of the gastric duplication cyst during the TEF repair in index

case underscores the importance of addressing all anomalies comprehensively to improve outcomes.

CONCLUSION(S)

This case emphasises the importance of early recognition, meticulous surgical planning and multidisciplinary management in neonates with complex congenital anomalies like TEF, pulmonary agenesis, and foregut duplications. Collaborative efforts among paediatric surgeons, neonatologists and paediatric specialists are essential for achieving favourable outcomes in such challenging cases. The clinician should evaluate thoroughly every TEF case to be prepared to encounter the other related congenital anomalies during the surgery.

REFERENCES

[1] Durkin N, De Coppi P. Anatomy and embryology of tracheo-esophageal fistula. In Seminars in Pediatric Surgery. 2022;31(6):151231. WB Saunders.

[2] Kayemba-Kay's S, Couvrat-Carcauzon V, Goua V, Podevin G, Marteau M, Sapin E, et al. Unilateral pulmonary agenesis: A report of four cases, two diagnosed antenatally and literature review. Pediatr Pulmonol. 2014;49(3):E96-102.

[3] Fukuoka S, Yamamura K, Nagata H, Toyomura D, Nagatomo Y, Eguchi Y, et al. Clinical outcomes of pulmonary agenesis: A systematic review of the literature. Pediatr Pulmonol. 2022;57(12):3060-68.

[4] Al-Salem AH, Kothari M, Oquaish M, Khogeer S, Desouky MS. Morbidity and mortality in esophageal atresia and tracheoesophageal fistula: A 20-year review. Ann Pediatr Surg. 2013;9(3):93-98.

[5] Kassif E, Weissbach T, Kushnir A, Shust-Barequet S, Elkan-Miller T, Mazkereth R, et al. Esophageal atresia and tracheoesophageal fistula: Prenatal sonographic manifestation from early to late pregnancy. Ultrasound Obstet Gynecol. 2021;58(1):92-98.

[6] Parelkar SV, Athawale HR, Sanghvi BV, Joshi PB, Kapadnis SP, Oak SN. Unilateral lung agenesis associated with esophageal atresia & tracheoesophageal fistula: Report of two cases & review of literature. J Pediatr Surg Case Rep. 2013;1(8):203-06.

[7] Patria MF, Ghislanzoni S, Macchini F, Lelii M, Mori A, Leva E, et al. Respiratory morbidity in children with repaired congenital esophageal atresia with or without tracheoesophageal fistula. Int J Environ Res Public Health. 2017;14(10):1136.

[8] Spataru RI, Popoiu MC, Ivanov M. Foregut duplication cyst associated with esophageal atresia-one-stage neonatal surgical repair. Indian J Surg. 2015;77(Suppl 1):52-55.

[9] Halilbasic A, Skokic F, Hotic N, Husaric E, Radoja G, Muratovic S, et al. Unilateral pulmonary agenesis and gastric duplication cyst: A rare association. Case Rep Pediatr. 2013;2013:608706.

[10] Kluth D, Fiegel H. The embryology of the foregut. In Seminars in Pediatric Surgery. 2003;12(1):03-09. WB Saunders.

PARTICULARS OF CONTRIBUTORS:

1. Additional Professor, Department of Paediatric Surgery, Post Graduate Institute of Child Health, Noida, Uttar Pradesh, India.
2. Associate Professor, Department of Paediatric Surgery, Post Graduate Institute of Child Health, Noida, Uttar Pradesh, India.
3. Assistant Professor, Department of Paediatric Surgery, Post Graduate Institute of Child Health, Noida, Uttar Pradesh, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Umesh B Singh,
Associate Professor, Department of Paediatric Surgery, Post Graduate Institute of Child Health, Noida-201310, Uttar Pradesh, India.
E-mail: singhub.2008@gmail.com

PLAGIARISM CHECKING METHODS: [Jain H et al.]

- Plagiarism X-checker: Nov 22, 2024
- Manual Googling: Apr 07, 2025
- iThenticate Software: Apr 17, 2025 (8%)

ETYMOLOGY: Author Origin

EMENDATIONS: 6

AUTHOR DECLARATION:

- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Nov 21, 2024**
Date of Peer Review: **Mar 05, 2025**
Date of Acceptance: **Apr 18, 2025**
Date of Publishing: **Sep 30, 2025**