

# Anorectal Malformations Associated with Esophageal Atresia in Neonates

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**Abstract:** *Tracheo- esophageal fistula (TEF) with anorectal malformation (ARM) is a recognized association while colonic agenesis is a rare cause of intestinal obstruction. We present our experience and search of literature in one such case of TEF with high ARM in addition to colonic agenesis in a newborn male child. This combination has not been reported.*

**Keywords:** Colonic Aggenesis; Tracheo- Esophageal Fistula; Anorectal Malformation

**Abbreviations:** TEF: Tracheo-Esophageal Fistula; ARM: Anorectal Malformation; EA: Oesophageal Atresia

## 1. Introduction

Anorectal malformations occur approximately in 1/1,500 to 1/5,000 live births. They may occur alone, but they can commonly have other associated anomalies or occur as a part of the combined anomaly. Furthermore, the problems of these associated anomalies could have more of an impact on the morbidity and mortality before and after surgical treatment. esophageal atresia with Tracheo- esophageal fistula (EA +TEF) is frequently associated with other congenital anomalies. The commonly associated systems are cardiovascular and gastrointestinal, including anorectal malformations. The associated anomalies adversely affect the outcome of the patient. Based on this association Waterson has given criteria for risk stratification and survival of neonates of EA+TEF.

## 2. Case Study

A one day male was admitted in NICU in SMIMER hospital with frothing and respiratory distress and no passage of stools. The patient was a full term normal delivery with a birth weight of 2.2kg and cried immediately after birth.

Antenatal history was not significant. On examination, the patient had an imperforate anus with poorly developed buttocks. A red rubber catheter or infant feeding tube could be passed orally only up to 8 cm from the alveolar margin. Radiographic studies confirmed the findings of EA with TEF (Figure 1). USG abdomen was done suggestive of faecal loaded large bowel loops Cross table prone X ray or invertogram was indicative of high ARM (Figure 2). Routine laboratories were within normal limits. USG skull and 2d echo was done to rule out congenital VACTREL anomalies, both were normal. Patient was admitted in NICU for invasive monitoring and kept on higher antibiotics and was planned for definitive surgical management. TEF repair was done via right thoracotomy extra pleural approach. On laparotomy, the dilated colon WITH colonic agenesis was found with the small bowel was completely developed up to the ileocaecal junction, and appendix was present. There was no dilatation of the proximal ileum and there was no enterovesical fistula. End transverse colostomy (Figure3) was performed however the patient succumbed to sepsis on postoperative day 3.



Figure 1: Esophageal Atresia with TEF

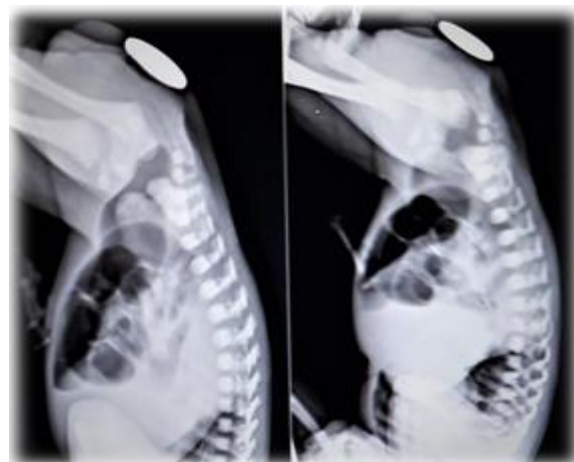
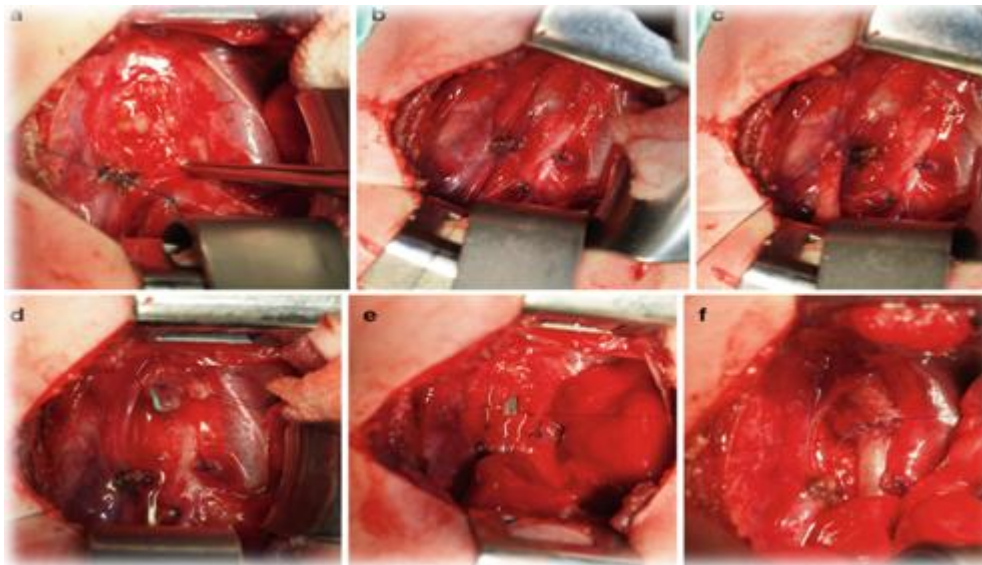


Figure 2: Invertogram Xray for Arm

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TEF with EA Repair Steps Via Right Thoracotomy



TEF Repair via Right Thoracotomy



Figure 3: Transverse Colostomy

### 3. Discussion

EA+TEF is associated with many congenital anomalies. The most frequently associated anomalies with EA±TEF are cardiac (49%) and anorectal malformations (15%). There have been many cases of TEF with ARM described. Colonic agenesis is a very rare cause of intestinal obstruction. The early disturbance in organogenesis that results in EA deformities, whatever the exact cause, also affects other organ systems. One theory that is proposed is of aborted hindgut. The exact embryogenesis is unknown development following obliteration of the inferior mesenteric artery early in fetal life.

The caecum and the right colon develop normally from the post axial midgut when this portion of the midgut is stimulated by normally developing hindgut. Thus improper development of the post axial midgut or presplenic gut is due to a primary disorder of the proximal end of the hindgut or post splenic gut. This case is the first reported case of the combination of EA+TEF with ARM and total colonic agenesis to the best of our knowledge. Although many factors may be involved in the occurrence of these

anomalies, it may be thought that anorectal malformations and esophageal atresia are types of midline defects, which present along the body. These defects occur as results of the combination of deficits in mesodermal migration and endodermal defects. On the other hand, in VACTERL association, there are two different explanations involving it; genetic factors and external environmental factors. First, the genetic factor model has been supported through animal experiments, and it proposes the formation of anomalies due to genetic mutations that induce signaling pathway abnormalities. Second, it is thought that external environmental factors such as maternal diabetes, hormonal exposure during infertility treatments, and exposure to toxic factors negatively influence the morphological development of the fetus, leading to malformations in the fetus.

In the management of a complex anomaly, the correlation with life support must be considered primarily in the neonatal period, and it is essential to do appropriately a staged procedure.

#### 4. Conclusion

Anorectal malformations associated with esophageal atresia, whether alone or as a part of a complex syndrome, had a relatively high frequency of associated anomalies in other organs, as well as a high mortality rate. It may be suggested that a staged and multidisciplinary approach is essential in the management of neonates with this anomaly. In addition, reasonable treatments considering these possibilities may lead to improved outcomes by preventing delays in the diagnosis of anorectal malformations associated with esophageal atresia, as well as the development of serious complications.

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