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Case Report

Esophageal Atresia, Tracheoesophageal Fistula, Congenital Heart Disease, Anorectal Malformation and Pyloric Atresia: A Case Report

Meriem Oumaya[1]*, Awatef Charieg[1], Chedi Saadi[1], Mariem Marzouki[1], Intissar Chibani[1], Yosra Ben Ahmed[1], Faouzi Nouira[1], Said Jlidi[1]

[1]Department of Pediatric Surgery B, Children Hospital Bechir Hamza, 1007, Tunis, Tunisia University of Tunis El Manar, Tunis, Tunisia

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Abstract: ***Introduction*:** Esophageal atresia with tracheoesophageal fistula (EA/TEF), anorectal malformation (ARM), and pyloric atresia (PA) are rare congenital anomalies. Their simultaneous occurrence, especially in association with complex cardiac defects such as double outlet right ventricle (DORV), is exceedingly rare. ***Case presentation*:** A male neonate was delivered at 33 weeks of gestation via emergency cesarean section due to labor dystocia. Prenatal ultrasound had only identified cardiac anomalies. At birth, the 2000-gram infant presented with respiratory distress. Clinical examination revealed excessive salivation, absence of an anal opening with no visible perineal fistula, and inability to pass an orogastric tube. Chest and abdominal radiographs demonstrated the orogastric tube coiled in the upper esophageal pouch and a single dilated gastric bubble without distal bowel gas, consistent with EA/TEF and suggestive of proximal intestinal obstruction. Echocardiography confirmed the presence of DORV. A right thoracotomy confirmed EA with a distal tracheoesophageal fistula, which was ligated. Given the neonate's hemodynamic instability, an esophageal anastomosis was deferred, and a staged approach was chosen, with cervical esophagostomy and feeding gastrostomy. During laparotomy, pyloric atresia with complete discontinuity between the stomach and duodenum was identified and managed with a gastro-duodenostomy. A colostomy was also performed for the high-type ARM. Despite initial postoperative stabilization, the infant deteriorated and died on day 15 from sepsis, persistent hypotension, and multi-organ failure. ***Conclusion*:** Although rare, patients with EA/TEF or ARM may present with associated pyloric atresia and complex cardiac anomalies. Early recognition and multidisciplinary approach are crucial for management of such challenging neonatal presentations.

Keywords: Congenital anomalies; Pyloric atresia; Case report.

INTRODUCTION

Esophageal atresia with tracheoesophageal fistula (EA/TEF), pyloric atresia (PA), and anorectal malformation (ARM) are rare conditions. Esophageal atresia (EA) with tracheoesophageal fistula (TEF) occurs in approximately 1 in 2,500–4,000 live births [1]. Anorectal malformations (ARM) are observed in approximately 1 in 5,000 live births. The incidence of intestinal atresia (IA) varies between 1.3 and 3.5 per 10,000 live births [2]. Congenital pyloric atresia (PA) is even rarer, occurring in approximately 1 in 100,000 newborns, and represents less than 1% of all intestinal atresia [3,4].

We report an exceptionally rare case of concurrent EA/TEF, PA, ARM, and complex cardiac anomalies in a single patient, underscoring the diagnostic and therapeutic challenges involved.

CASE REPORT

A male neonate was delivered at 33 weeks of gestation via emergency cesarean section due to labor dystocia. Prenatal ultrasound had only identified cardiac anomalies. At birth, the 2000-gram infant presented with respiratory distress. His Apgar scores were 7 and 8 at 1 and 5 minutes, respectively. He was admitted to the neonatal intensive care unit (NICU) and required intubation and mechanical ventilation.

Clinical examination revealed excessive salivation, absence of an anal opening with no visible perineal fistula (Figure 1), and failure to pass an orogastric tube. Chest and abdominal X-ray

***Corresponding Author:** Meriem Oumaya
Department of Pediatric Surgery B, Children Hospital Bechir Hamza, 1007, Tunis, Tunisia University of Tunis El Manar, Tunis, Tunisia

demonstrated the orogastric tube coiled in the upper esophageal pouch. The stomach appeared dilated and air-filled, with no distal bowel gas, findings consistent with the presence of esophageal atresia with a distal tracheoesophageal fistula and suggestive of proximal intestinal obstruction. Echocardiography confirmed the presence of a double outlet right ventricle (DORV).

A right thoracotomy confirmed EA with a distal tracheoesophageal fistula, which was ligated. Given the neonate's hemodynamic instability, an esophageal anastomosis was deferred, and a staged approach was chosen, with cervical esophagostomy and feeding gastrostomy.

During laparotomy, pyloric atresia with complete discontinuity between the stomach and duodenum was identified (Figures 2, 3) and managed with gastro-duodenostomy. A left-sided double-barrel sigmoid colostomy was also performed for the high-type ARM, based on the absence of a perineal fistula and anal opening.

The immediate postoperative course was uneventful. The colostomy became functional on third postoperative day, and enteral feeding via the gastrostomy tube was initiated on the fifth postoperative day. However, on day 15, the infant developed severe sepsis, which progressed to multiple organ dysfunction syndrome and death.

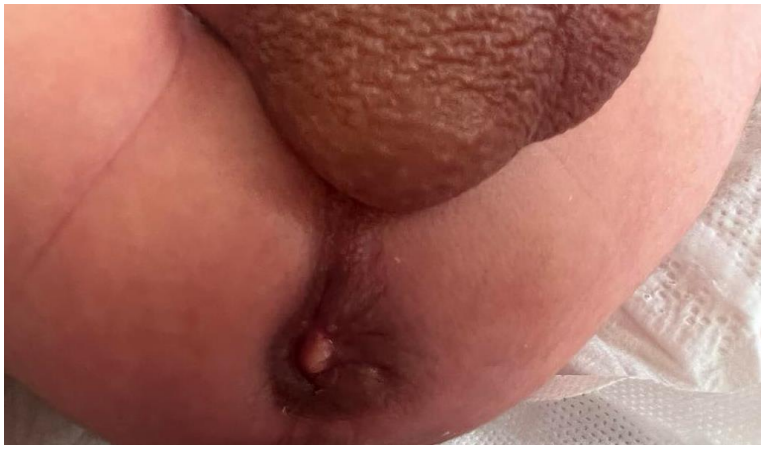

Figure 1: Imperforate Anus

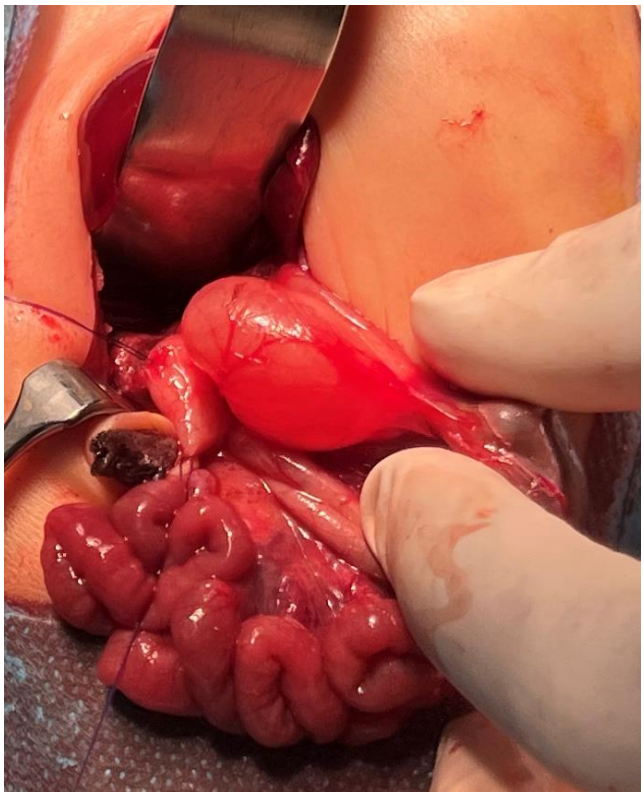

Figure 2 : Peroperative View Showing a Dilated Stomach, an Atretic Pylorus and a Small Duodenum

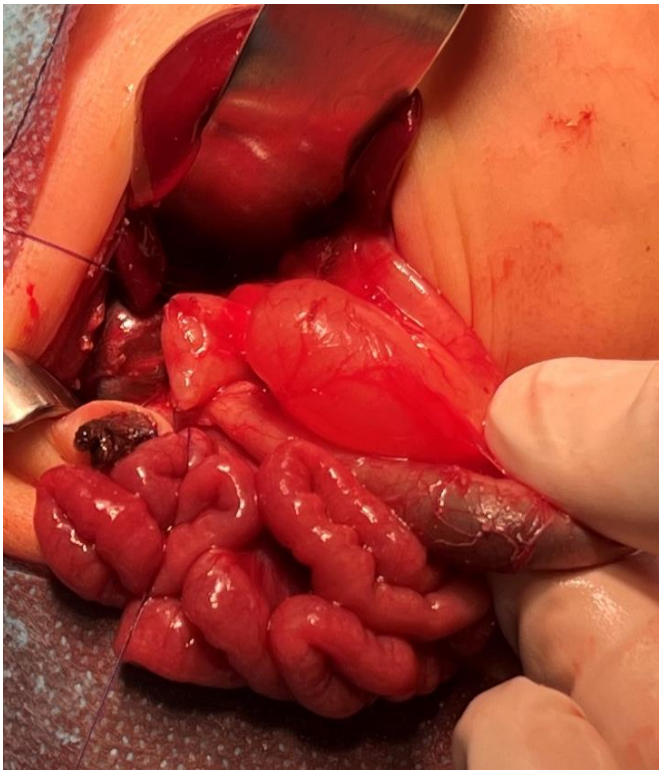

Figure 3: Agap Between the Stomach and the Entire Duodenum Representing PA Type3

DISCUSSION

The association of multiple congenital anomalies is rare. Previous reports have described cases of duodenal atresia, malrotation, segmental dilatation of the colon, and ARM [5], as well as association of EA, duodenal atresia, biliary atresia, and pancreatic duct atresia [6]. Cases involving EA/TEF, duodenal atresia, malrotation, and ARM have also been documented [7]. Our case describes a male neonate with an exceptionally rare constellation of five distinct congenital anomalies: esophageal atresia with tracheoesophageal fistula (EA-TEF), pyloric atresia (PA), double outlet right ventricle (DORV) and anorectal malformation (ARM). To the best of our knowledge, no previous reports have documented the simultaneous occurrence of these five congenital anomalies in the same patient

This association of congenital anomalies poses significant diagnostic and therapeutic challenges, emphasizing the importance of early prenatal diagnosis in managing such complex cases. While ultrasound is less effective for identifying ARM, it is crucial for diagnosing intestinal atresia. In the second trimester, prenatal ultrasound may suggest PA in the presence of polyhydramnios and a dilated stomach [8, 9]. In this case, fetal ultrasound identified only cardiac abnormalities, underscoring the need for comprehensive antenatal screening to detect additional anomalies. Earlier detection, ideally in the first and second trimesters, could have significantly influenced perinatal management and more informed decision-making [10].

Managing such a complex case requires a multidisciplinary approach. The infant deteriorated rapidly post-delivery, necessitating an immediate admission to the neonatal intensive care unit. The diagnosis of EA/TEF was based on clinical signs and confirmed by radiographic imaging. The presence of an imperforate anus further complicated the clinical picture, and the unexpected discovery of pyloric atresia during laparotomy added another layer of complexity to the surgical interventions [9].

There are three recognized anatomic variations of PA: Type 1 is a pyloric membrane or diaphragm, which can be multiple. This is the most common type, reported in 57% of patients. In type 2, the pyloric canal is replaced with a solid core of tissue (34% of cases) and type 3 is an atretic pylorus with a gap between stomach and duodenum [11]. It is the least frequent (9%) and was the case of our patient. A gastro-duodenostomy was performed to restore intestinal continuity [12].

The differential diagnosis between pyloric atresia (PA) and duodenal atresia (DA) can be challenging. However, in our case, several intraoperative findings were characteristic of type 3 PA. A complete anatomical discontinuity between the stomach and the duodenum was clearly observed, with no identifiable pyloric segment. The entire duodenum was collapsed, while the stomach was markedly dilated. In contrast, in duodenal atresia, the pylorus is typically patent and the proximal duodenum is dilated, features that were not observed in our case. Furthermore, duodenal atresia is almost exclusively located in the second portion of the

duodenum (D2), either preampullary or postampullary. Involvement of the first portion of the duodenum (D1) is exceedingly rare. Reports of duodenal atresia involving the first portion (D1) are exceedingly rare. Even in such exceptional case, a short segment of proximal duodenum is usually identified before the site of atresia, often associated with annular pancreas [13].

Therefore, the absence of a recognizable pyloric segment and the presence of a collapsed duodenum with a clear gap between the stomach and duodenum strongly support the diagnosis of Type 3 pyloric atresia rather than duodenal atresia [3, 4, 9, 11, 12].

The management of such complex congenital anomalies is challenging and requires collaboration among multiple specialties, including pediatric surgery, neonatology, anesthesiology, and pediatric cardiology [14, 15, 16]. A well-coordinated healthcare team is essential to optimize patient outcomes [4, 17, 18].

Despite intensive medical interventions, the neonate developed severe sepsis on the 15th postoperative day, leading to multiple organ failure and death. This tragic outcome highlights the challenges of treating complex congenital anomalies, particularly in resource-limited countries. It also reinforces the importance of prenatal diagnosis in improving management strategies for such rare and intricate cases.

Studies conducted in developing countries have assessed pregnant women's knowledge of congenital anomalies. Through our study, we further emphasize the need for public health programs to improve awareness and promote early detection of these malformations [19, 20]. Genetic counseling for families at risk of congenital anomalies has also been recommended.

In light of this exceptional case, several recommendations can be made to enhance clinical practice. First, use of genetic counseling with early and comprehensive antenatal screening should be advocated for all pregnancies at risk for congenital anomalies. Second, the importance of a multidisciplinary team approach cannot be overstated, as these cases need close collaboration among specialists. Finally, increased awareness and education among healthcare providers about the potential co-occurrence of rare congenital anomalies can facilitate prompt diagnosis and intervention.

CONCLUSION

Although rare, patients with EA/TEF or ARM may present with associated pyloric atresia and complex cardiac anomalies. Early recognition and a multidisciplinary approach are essential for optimizing perinatal management and surgical planning.

Informed Consent:

Consent to publish this case report was not obtained. The report does not contain any personal information that leads to the identification of the patient.

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Declaration of Competing Interest:

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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