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Esophageal atresia associating gastrointestinal malformations: a study of clinical approach

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ABSTRACT



Digestive tract malformations requiring surgical repair in association with esophageal atresia are rare occurrences. Because of this uncommon presentation of esophageal atresia, its evaluation and management are often difficult, requiring extensive workup and multiple surgical procedures. We present our experience with esophageal atresia associating gastrointestinal congenital anomalies in the last 10 years. Clinical and surgical perspectives were pointed willing to make relevant observations in matters of diagnosis and treatment strategy in these patients. Therefore, 7 cases resembling this pattern were identified – duodenal atresia and anorectal malformation being the most common coexistent malformations. All the cases exhibited technical and procedural staging difficulties that we shaped according to the patient's general picture in the absence of a standardized approach. Careful preoperative follow-up, multidisciplinary communication, watchful waiting and an individually patient-shaped approach are elementary requirements for good outcomes in complex cases of EA.

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Introduction

Esophageal atresia (EA) occurs in 1:2500 births and it is associated with other malformations in about half of the cases, out of which the most common are: cardiac (29%), anorectal (14%), genitourinary (14%) and gastrointestinal (13%) congenital anomalies. Most cases of EA are sporadic and only 1% constitute into syndromes [1]. The incidence of duodenal atresia (DA) and anorectal malformations (ARM) is 1-2:10000 births [2], and 2-6:10000 births respectively [3]. About 8-11% of EA cases are complicated by association with ARM [4] and 6% of EA patients present with concomitant DA [5]. There are also extremely rare digestive malformations reported in conjunction with EA, such as foregut duplication cysts [6] or microgastria [7].

The risk factors for esophageal atresia are still a subject of research. Environmental factors, such as maternal alcohol and smoking abuse, toxic exposure, infections and genetic conditions were considered [8-10]. In a study by

Melek et al., serum Mg, Mn, Zn, Cd, Cu levels were significantly higher in newborns with EA than in healthy newborns, suggesting that some minerals and trace elements may be involved in the biomolecular mechanisms of this disease [11].

Until the early '90s, when the new Spitz et al. revision over the EA risk-groups was published, the coexistence of other congenital anomalies with EA was considered a poor prognostic factor for the outcomes of these newborns [12]. Since then, significant progress in neonatal intensive care has been made, allowing only severe cardiac anomalies and lower birthweight (BW) to be fearsome for the outcome of EA patients [13].

However, uncommon presentations of esophageal atresia associating gastrointestinal malformations requiring surgical approach are reported in isolated cases or small case series and they may raise difficulties in matter of approaching and staging the procedures. This situation may not be standardized; therefore, we consider that literature refinement by varied experience is needed [14,15].

Materials and Methods

The medical records of newborns operated on for EA by a unique surgical team in the Emergency Clinical Hospital for Children “Marie S. Curie” in Bucharest in the last 10 years (2011-2020) were reviewed, out of which the cases associating digestive malformations requiring surgery were selected. The cases were considered from a clinical point of view willing to make relevant observations in the approach of these particular EA situations. Clinical and diagnostic aspects, surgical timelines and outcomes were outlined, analyzed and discussed aiming at highlighting the comprehensive diagnostic strategy and operative planning done by multidisciplinary teams.

Table 1. The summary of EA cases associating congenital anomalies of the digestive tract requiring surgery.

	EA classif.	Sex	BW	GA	Prenatal diagnosis	Associated gastrointestinal malformation
1	Type C	F	2,820	40	No	Esophageal duplication cyst
2		M	1,100	34	No prenatal monitoring	Duodenal atresia
3		M	2,830	37	No prenatal monitoring	Duodenal atresia
4		M	2,600	33	Polyhydramnios	Duodenal atresia
5		M	1,580	36	Polyhydramnios	Duodenal atresia + Perineal fistula
6		M	1,700	35	Polyhydramnios	Imperforate anus without fistula
7		F	2,620	38	Polyhydramnios Absence of stomach bubble	Congenital microgastria

Seven cases of EA associating surgical congenital anomalies of the gastrointestinal tract were reviewed and summarized in Table 1. The informed consent for surgery was obtained in all the cases from the parents as legal representatives, after a comprehensive discussion about the risks, benefits and therapeutic alternatives, within the limits of reasonable disclosure [16]. Safe surgery checklist for pediatric patients was used in all cases as a tool of avoiding preventable postoperative complications and increase family satisfaction related to the medical care [17-19]. The male: female ratio is 5:2. Their mean birthweight (BW) is 2,178 g and the mean gestational age in weeks (GA) is 36. Two cases did not present prenatal monitoring. The five remaining cases were monitored ante-partum, out of which one did not reveal any sign of fetal bowel

obstruction, three associated polyhydramnios and in one case, stomach bubble could not be identified together with the occurrence of polyhydramnios. All cases had type C EA according to Gross classification and in all cases EA repair was done via right open thoracic approach. In all cases, an informed consent was signed by the legal representative prior to the surgical treatment.

Results

EA associating esophageal duplication cyst: case 1

The first patient is a female newborn delivered at 40 weeks of gestation, with a BW of 2,820 g. No prenatal diagnostic signs were observed. Failure to pass a nasogastric tube shortly after birth presumed the diagnosis of EA, therefore she was transferred to our unit, where type C EA was outlined by barium swallowing and plain abdominal X-ray (upper pouch ending at the 3rd thoracic vertebra and the presence of gaseous distension of the stomach and of the small bowel). The surgical intervention was performed in the first day of life via right thoracotomy, being an extra-pleural approach. Intraoperative findings confirmed the diagnosis with an approximately 2 cm gap between the two esophageal endings. Additionally, a well-defined cystic lesion was identified closely attached to the distal pouch non-communicating with the esophageal lumen, developed in its muscular layer, adjacent to the tracheo-esophageal fistula (TEF) (Figure 1). The lesion was excised and end-to-end esophageal anastomosis was done. The postoperative evolution was uneventful [6].

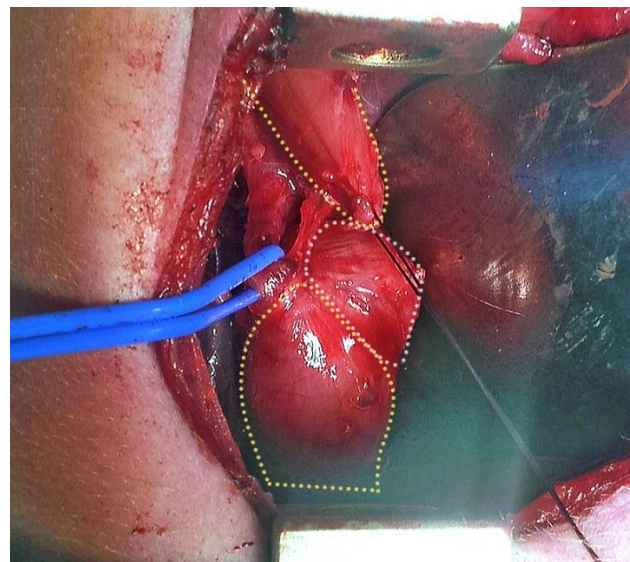


Figure 1. Foregut duplication cyst (white dotted stroke) and the two esophageal ends (yellow dotted stroke). The TEF is isolated on the blue rubber loop.

EA associating DA exclusively: cases 2, 3, 4

The second case is a 1,100 g male, born at a GA of 34 weeks. The pregnancy was not medically monitored. Failure to pass a nasogastric tube and respiratory distress

referred the case to our unit, where the diagnosis of type C EA and DA was made – the esophagogram and the plain abdominal aspect of the X-ray showed upper pouch ending at the 3rd thoracic vertebra, the presence of gaseous distension of the stomach, but not of the small bowel (Figure 2).



Figure 2. Type C EA associating duodenal atresia preoperative barium swallowing, full body X-ray.

The newborn was operated on in the first day of life: ligation of the TEF and end-to-end esophageal anastomosis were performed. The postoperative evolution was slowed down due to severe respiratory distress syndrome, but the absence of gas below the duodenal bulb persisted on subsequential plain X-rays prior to the esophageal anastomosis, together with high volumes of nasogastric aspirate. A transverse abdominal open approach in the 14th day of life and diamond-shaped duodenoduodenostomy were performed. The postoperative evolution was uneventful.

The third and fourth cases had a similar clinical course. One was not prenatally monitored, while the other presented polyhydramnios before birth. In both cases, DA was suspected due to the absence of intestinal gaseous distension beyond the duodenal bulb. The patients were operated on in the first, respectively the 2nd day of life; ligation of the TEF and end-to-end esophageal anastomosis and duodenoduodenostomy were performed early in the 3rd, respectively the 5th day of life, when anesthetic support was allowed safely. In both cases, the postoperative evolution was uneventful.

EA associating DA and perineal fistula: case 5

A 1,580 g male born at 36 weeks of pregnancy was transferred to our unit. EA was suspected (failure to pass a nasogastric tube and severe respiratory distress). The case was associated with perineal fistula and no meconium passage. The esophagogram and the plain abdominal X-ray determined the association of EA with DA in the same manner as in the previous cases. The surgical repair of the malformation complex was staged in: TEF ligation and primary anastomosis of the esophagus (in the first day of life) and concomitant duodenostomy and Dennis-Browne proctoplasty (in the 14th day of life) which could be delayed because the fistula allowed catheterization and colon decompression through repeated enemas. The postoperative evolution was uneventful.

EA associating imperforate anus without fistula: case 6

A 1,700 g male born at 35 weeks of GA was transferred to our clinic for the evaluation and management of EA and imperforate anus. He was under severe respiratory distress and the required intubation procedure was difficult to perform. EA was confirmed by the esophagogram and he first underwent surgery in the 4th day of life - distal TEF ligation, Stamm gastrostomy (primary esophageal anastomosis could not be achieved because of the long gap between the two pouches) and terminal colostomy with mucous fistula. The CT done at 40 days of life suspected congenital subglottic tracheal stenosis, which was later confirmed by bronchoscopy in the 49th day of life, when tracheostomy was subsequently done. In the 67th day of life, cervical esophagostomy was performed. The patient was discharged after 76 days of hospitalization. At 4 months of age, posterior sagittal anorectoplasty was performed and – 4 months later, after several anal dilatations for anal stenosis – stoma reversal was done. At 14 months of age, tracheoplasty was done and, currently, the patient is scheduled for transverse colon esophagoplasty at 16 months.

EA associating congenital microgastria: case 7

A full-term 2,620 female newborn was transferred to our unit with a high suspicion of EA, later confirmed by the esophagogram. Since the hospitalization took place during the COVID-19 pandemic, all regulations were observed to prevent the intrahospital transmission of the infection: PCR testing upon admission, rigorous disinfection of the surfaces, adequate protective personal equipment of the medical personnel [20-22]. She underwent surgery in the 2nd day of life, wherein TEF ligation was done, but esophageal anastomosis could not be achieved because of the long gap between the two ends. Therefore, Stamm gastrostomy was approached revealing a short (approx. 15 mm), thin tube-shaped stomach – congenital microgastria. Gastrostomy was achieved with technical difficulty. The postoperative evolution was slow

and difficult in matters of enteral feeding because of the extremely small volume stomach. The barium gastrography done at 2 months through gastrostomy revealed signs of stomach enlargement (Figure 3).



Figure 3. Barium gastrogram through a gastric catheter 2 months after TEF ligation, showing signs of gastric enlargement and gastro-esophageal reflux on the distal esophagus ending.

Beginning with 3 months of life, the patient begins to exhibit isolated respiratory distress episodes with feeding times. Methylene blue, administered during gastrostomy, was consequently expelled in the cough sputum. At 5 months of life, right thoracotomy was performed again and ligation of a eso-bronchial fistula was done together with end-to-end delayed primary esophageal anastomosis. The patient developed severe gastroesophageal reflux disease and recurrent esophageal stricture. Re-do esophageal anastomosis was done at 9 months of age, after no response at esophageal dilations. At 12 months of age, gastric augmentation through Hunt Lawrence procedure was performed. Since then, recurrent esophageal stricture persisted in the context of gastroesophageal reflux disease, but the patient had a favorable outcome throughout endoscopic dilatations. The patient is currently 5 years old, weighing 16 kilos, being well-integrated into the schooling system, and returns for endoscopic check-ups every 3-6 months. The patient presents persistent GERD and sliding hiatal hernia which are currently managed conservatively.

Discussions

All our cases exhibited challenging situations in matters of clinical or surgical approach. A good preoperative preparation through a multidisciplinary

approach is important to prevent postoperative complications [23-26].

In the first case, the esophageal duplication cyst in close contact with the distal esophageal pouch and TEF in the muscular layer endangered the integrity of the esophageal wall. By increasing the gap distance between the esophageal ends through a disregarding breach or ischemia in the esophageal wall, the segmented resection of the esophagus could have led to the indication of delayed primary esophageal anastomosis and gastrostomy. The careful dissection of the cyst and its non-communication with the esophageal lumen feature made tension-free anastomosis possible at birth. Foregut duplication cysts are uncommon findings in neonates and infants. They become even more particular when located in the mediastinum and associate heterotopic pancreatic tissue [27].

All our EA associating duodenal malformations could be diagnosed early without additional unnecessary workup. This was possible because they were either type 2 or type 3 DA in conjunction with type C EA, therefore the gaseous distension is limited to the stomach and duodenal bulb. In the fifth case, in which the perineal fistula complicates the congenital malformation association, colostomy was avoided by repeated decompression enemas and Dennis-Browne proctoplasty was the procedure of choice concomitant with the duodenal repair. The patient is now 5 years old, presenting normal bowel movements, no constipation, nor soiling.

DA may be recognized on plain X-rays if distal TEF is present or later, when esophageal continuity is established. Staged surgery is beneficial for the baby because it allows respiratory distress and metabolic stabilization after birth, especially in preterm babies [5,14]. Gastro-esophageal reflux disease is the most frequent complication of EA associated with DA because of the esophageal dysmotility together with duodeno-gastric reflux. This may lead to early anastomotic strictures and gastro-esophageal reflux disease related complications. Therefore, we think duodenal repair should be done as soon as the patient allows safe surgical procedure under safe conditions. This will also avoid unnecessary prolonged total parenteral nutrition [28]. In our opinion, early postoperative acquiring detailed dynamic barium swallow images in a child presenting gastro-esophageal reflux after primary EA repair will help avoid subsequent delayed diagnosis of more slippery duodenal malformation-like webs.

The sixth case required two concomitant surgical procedures: TEF ligation and colon decompression. Esophagostomy was considered, taking into account the safety of the long-term further operative plan and the coexistent tracheal malformation. Wide-spaced staged procedure allowed the stabilization of the patient's respiratory status, but prolonged intubation required long-term antibiotic therapy, thus increasing the susceptibility to various infections. Whenever necessary, we prefer to

perform esophageal substitution with transverse colon graft, passed retrosternally. When this route is not available (especially when the baby underwent previously cardiac surgery or requires surgery) we place the colonic graft into the posterior mediastinum. We uphold this technique in long gap esophageal atresia because of our good experience with pediatric colon pathology [29], but, first of all, because our postoperative results are satisfactory.

Congenital microgastria in the seventh case led to a series of successive morbidities. Initially, the operative technique for Stamm gastrostomy was highly difficult because of the gastric volume and size which did not allow filling the balloon of the gastric catheter and gastric dislodgement and attaching it to the anterior wall of the abdomen. Furthermore, low-volume continuous combined oral and gastrostomy feeding were performed, but the patient failed to thrive and presented multiple episodes of aspiration pneumonia related to high gastroesophageal reflux, which also contributed to refractory esophageal anastomotic stricture.

V.A.C.T.E.R.L. association may be defined in up to half of the EA cases associating other malformations. This comprises fewer manifesting conditions such as vertebral anomalies [30]. However, in a recent comprehensive study, Amelot et al. [31] proposes a redefinition of V.A.C.T.E.R.L. association by including spinal defects, which may lead to a noteworthy turn into long term multidisciplinary management of these patients who might associate fecal or urine dynamics conditions. Several authors showed that infants born with long-gap esophageal atresia (LGEA) may demonstrate various neurological disorders, so they should be closely monitored for further signs of delayed motor or cognitive development and associated complications [32-35].

Highlights

- ✓ Esophageal atresia/tracheal esophageal fistula is a rare congenital anomaly.
- ✓ Multidisciplinary approaches in matters of safety and timing are essential features when the operative plan is tailored on atypical congenital malformation complexes and clinical situations which cannot be standardized by treatment protocols.

Conclusions

EA needs to be carefully managed in careful high-end neonatal intensive care centers. This will allow the early and accurate diagnosis of the spectrum of associated anomalies. The multidisciplinary communication in matters of safety and timing are essential features when the operative plan is tailored on atypical congenital malformation complexes and clinical situations which cannot be standardized in terms of treatment protocols.

Long term good outcomes in difficult EA cases come from a well-staged approach where each surgical step is set up according to the patient's immediate needs in terms of survival and long-term morbidities.

Conflict of interest disclosure

There are no known conflicts of interest in the publication of this article. The manuscript was read and approved by all authors.

Compliance with ethical standards

The patients' consent was obtained. All patients admitted to "Marie S. Curie" Emergency Clinical Hospital for Children Bucharest systematically sign a consent of agreement that their anonymous clinical data may be used for scientific and educational purposes.

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