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CASE REPORT

Esophageal Atresia with positive family history: A case report

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Esophageal Atresia, Tracheoesophageal Fistula, Congenital Abnormalities

Abstract

Introduction: Esophageal atresia is a malformation marked by discontinuity of the esophagus, where the proximal part ends in a cul-de-sac, with or without the presence of a fistula with the trachea. An incidence pathology of 1 in every 2500 to 4500 live births, predominantly in individuals of white race and males, whose diagnosis can be made in the prenatal period by ultrasound or in the post-natal period when the clinical suspicion is present. The diagnosis must be early for treatment success, avoiding complications such as the evolution from pneumonia to respiratory failure. In view of the investigation, family history can be an alert for early suspicion. **Objectives:** To describe a case of esophageal atresia with distal tracheoesophageal fistula associated with heart disease, diagnosed after birth, in a female child, who had a positive family history for this condition, in the absence of a diagnosis of known genetic syndromes. **Method:** This is a descriptive observational study with data collection from the patients record at the Maternity Hospital. Literature reviews were carried out on platforms such as Lilacs, Scielo, Pubmed and research in Pediatric and Pediatric Surgery books on the subject. **Conclusions:** In the reported case, the diagnosis occurred after birth, with the newborn being referred early for surgical evaluation, which presented a successful approach, without complications. We hope that with the report, it will be possible to establish the family history as a point of the anamnesis to be valued in the investigation of the newborn.

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INTRODUCTION

Esophageal atresia (EA)with or without tracheoesophageal fistula corresponds to a complex of congenital anomalies characterized by incomplete formation of the esophagus and can occur with or without abnormal communication between it and the trachea[1]. Its prevalence is 1 in every 2500 to 4500 live births, predominantly in white and male individuals[2,3]. It has a higher incidence in twins, in children of older mothers and in primiparity. It is also frequently associated with chromosomal anomalies[4].

The exact etiology is unknown. It is supposed to be an anomaly that occurs during the process of division of the foregut, or primitive intestine, during the fourth and fifth week of embryonic development[5]. There is an interruption in the esophageal tubular formation, or an incomplete separation between the two portions of the foregut, resulting in esophageal atresia, with or without tracheoesophageal fistula[5,6]. Some lines of research investigate changes in neuropeptides in the process of esophageal formation, and others highlight environmental factors such as maternal use of dipyrone, exogenous sex hormones, maternal smoking and alcoholism, gestational diabetes, and maternal infections[3]. Other factors such as intrauterine anoxia or stress could cause local vascular impairment, resulting in focal esophageal necrosis. There is also a strong association with other chromosomal or structural anomalies, such as trisomy 18, VACTERL Syndrome and CHARGE[7] Syndrome.

Esophageal atresia (EA) is classified according to its location and whether or not there is an associated fistula. These clinical variants are: isolated EA without fistula (8% of cases); EA with proximal fistula (1% of cases); EA with distal fistula, the most common, (87% of cases); EA with proximal and distal fistula (1% of cases); Isolated tracheoesophageal fistula, without EA (4% of cases)[8]. The diagnosis may be suspected in utero when prenatal ultrasonography demonstrates the absence of a gastric bubble, dilatation of the proximal esophageal stump, absence of fluid in the fetal intestine and, mainly, polyhydramnios[3].The postnatal clinical picture is characterized by difficulty in swallowing of saliva and milk, increased foamy secretion that accumulates in the esophageal cul-de-sac, exteriorizing through the mouth and nostrils. The secretion can be aspirated through the airways, causing dyspnea and cyanosis, potentially complicating with aspiration pneumonia. When trying to pass an oral or nasogastric tube, there is difficulty in progressing, progressing to about 10 to 12 cm[9].

Chest X-rays can reveal the esophageal proximal end distended with air, the bending of the gastric tube, and the abdominal X-ray shows the presence of air in the gastrointestinal tract, which would be an indication of a distal fistula[1]. After confirming the diagnosis, other malformations should be screened through total abdominal ultrasound and transthoracic echocardiography, as some studies claim that the congenital disorders most associated with atresia are those related to the cardiovascular system[3].

Surgical correction of the esophagus can be performed in a single step or divided into 2 steps, depending on the type and extent of the anomaly and the clinical condition of the newborn[3]. With advances in surgical treatment, mortality has drastically decreased, with survival of up to 95%. There is a high possibility of cure, but a quick diagnosis is necessary for adequate treatment and reduction of complications[10].

Due to the importance of the subject and the rarity of reports of esophageal atresia with family kinship, we chose to report the case of a newborn, female, with a diagnosis of esophageal atresia with the presence of a distal esophageal tracheal fistula associated with heart disease, in which the patient had a positive family history.

CASE REPORT

Caucasian newborn (NB), female, born by cesarean section, gestational age of 40 weeks and 6 days, according to the first trimester ultrasound (12 weeks and 4 days), APGAR scores 9 and 10, water rupture during the delivery, meconium in the amniotic fluid, without the need for resuscitation. Intrapartum prophylaxis was performed with ampicillin due to a positive urine culture for streptococcus agalactiae (GBS). Birth weight of 3275 grams, suitable for gestational age, length and head circumference also suitable. Negative maternal serologies in the first and third trimester. Mother denied comorbidities, reported not having used alcohol or other drugs during pregnancy. Morphological ultrasound without changes, according to the report.

The patient's father was born with esophageal atresia with tracheobronchial fistula, operated on in the neonatal period, with no history of other malformations. However, he had several post-surgical complications, requiring care in the intensive care unit for a prolonged period. No family history of other malformations.

After birth, the NB was in good condition. General examination, physical examination, cardiac and pulmonary auscultation and abdominal palpation showed no abnormalities. The NB evolved with the exteriorization of a large amount of aerated salivation, which obstructed the upper airways, resulting in cyanosis. Secretion aspiration was performed with improvement in saturation without the need for oxygen therapy, but salivation persisted. She maintained O2 saturation between 94 and 98%, with no signs of respiratory distress, vesicular murmur present bilaterally, presence of diffuse rhonchi and well perfused extremities. Segmental physical examination without other abnormalities. An attempt was made to use an orogastric tube to perform gastric lavage, with difficulty in progressing. Fasting, serum therapy, chest x-ray and general exams were ordered.

The NB evolved with repeated drops in saturation, vomiting and nausea, which improved after aspiration of the upper airway. In the chest x-ray, the positioning of the probe in the esophageal stump (bent) at the level of the T2 vertebra, expanded lungs, presence of air in the stomach and intestine was identified.

The patient was referred to the neonatal ICU for further investigation and follow-up, where another contrast-enhanced x-ray was performed (Figure 1), confirming the diagnosis of esophageal atresia with tracheoesophageal fistula.



Figure 1. Contrast-enhanced chest X-ray demonstrating blind esophagus and tracheoesophageal fistula.

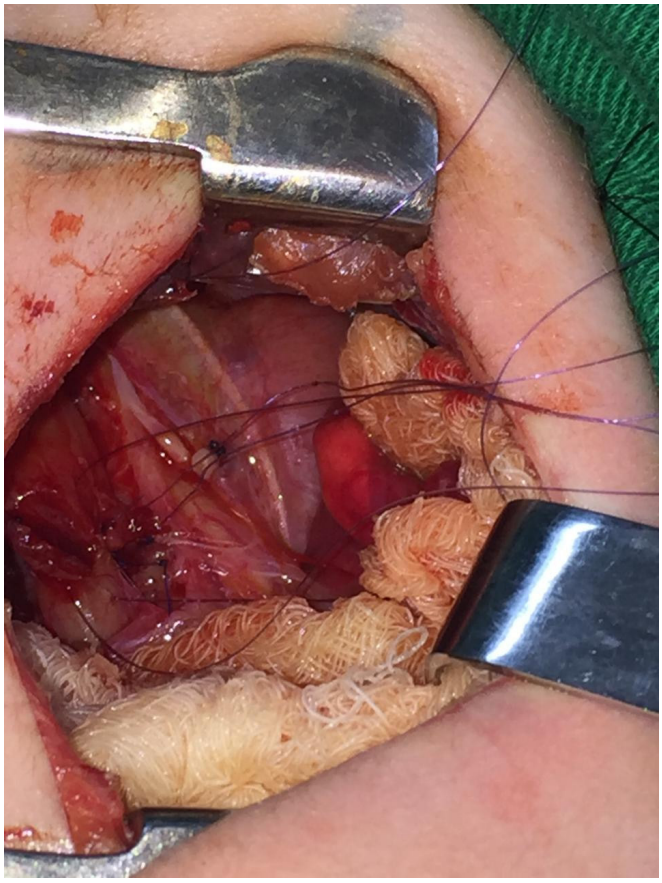


Figure 2. Photos of the surgical correction: primary end-to-end anastomosis of the esophagus, in a single plane, through a right thoracotomy.

Preoperative tests, total abdominal ultrasound and echocardiography were ordered to rule out the presence of other malformations. The echocardiogram showed an ostium secundum-type atrial septal defect with primary pulmonary hypertension, without clinical repercussions. Surgical correction was performed (figure 2) on the 5th day of life of the patient, with end-to-end primary anastomosis of the esophagus through a right thoracostomy.

Due to the good evolution of the condition, on the ninth postoperative day, a chest X-ray was performed with oral contrast, showing no abnormalities. Newborns remained in the ICU until the 15th day of life, remained in the ward for another 6 days and were discharged on the 21st day of life.

DISCUSSION

This report presents the case of a newborn (NB) with esophageal atresia and distal tracheoesophageal fistula, not diagnosed during prenatal care. The morphological ultrasonography performed during the gestational period did not show alterations such as the absence of a gastric bubble, dilation of the esophageal stump or the presence of polyhydramnios, thus, an intrauterine diagnosis was not possible. Most cases of esophageal atresia are diagnosed after breastfeeding (as in the case in question) when presenting regurgitation, cyanosis after breastfeeding, excessive and aerated salivation. Therefore, to avoid complications, it is extremely important that the diagnosis be made early.

Esophageal atresia predominates in males[2], the patient presented in this case was female, that is, an uncommon case. The incidence of EA increases in mothers with advanced age and in primiparity[4], the mother of this NB was primiparous, however the mother was young.

The patient in the case presented an echocardiogram showing congenital heart disease, thus demonstrating the importance of performing an echocardiography to investigate associated heart diseases, since these pathologies are frequent in cases of esophageal atresia.

The uniqueness of this case is due to the presence of a family history, in this case, the patient's father had presented the same condition at birth. A positive family history may be related to a genetic disease with a hereditary characteristic[11]. When esophageal atresia with tracheoesophageal fistula occurs as a feature of a genetic syndrome or chromosomal abnormality, it can be grouped into families according to the inheritance pattern of this condition[12].

Esophageal atresia can be clinically divided into isolated and syndromic forms. The etiologies of syndromic EA are multifactorial, being associated with environmental factors, chromosomal anomalies, malformations, genetic mutation, genetic disorders. However, there is little information about the causes of isolated EA. It is suspected that maternal exposure to environmental factors is responsible, such as the use of statins, tobacco, exogenous sex hormones or work in agriculture or horticulture[13]. In this case report, the mother denied any exposure to these factors. Other studies have shown that in EA, first-degree relatives are more likely to have VACTERL[2] spectrum malformations. The father of the NB in this case had esophageal atresia, without having developed other anomalies of the VACTERL complex.

CONCLUSION

A family history of EA, not associated with genetic syndromes, remains an uncommon event, both in the literature and in clinical practice. The success of the approaches will provide a new generation of children whose parents were adequately treated in childhood. The family history of EA should always be investigated and the physical examination carried out completely, since it is related to other genetic syndromes associated or not with heredity. Questioning the parents about the presence of malformations in the family can point early to a more specific investigation of the newborn, and thus the diagnosis is made early, which allows surgical correction, reducing the number of complications, sequelae and increasing life expectancy.

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