CASE REPORT

UDC: 616-053.32-007-089:617.55
https://doi.org/10.2298/VSP170322135S

Congenital diaphragmatic hernia associated with esophageal atresia, tracheoesophageal fistula and total anomalous pulmonary venous connection in a premature twin newborn

Kongenitalna dijafragmalna kila udružena sa atrezijom jednjaka, traheoezofagealnom fistulom i totalnim anomalnim utokom plućnih vena kod prevremeno rođenog blizanačkog novorođenčeta

Djordje Savić*†, Blagoje Grujić*†, Nikola Stanković*, Maja Miličković*†, Zoran Stanković*, Vladimir Kojović*†

***Mother And Child Health Care Institute of Serbia „Dr Vukan Čupić“, Belgrade, Serbia; University of Belgrade, †Faculty of Medicine, Belgrade, Serbia**

Abstract

Introduction. Congenital diaphragmatic hernia (CDH) with concomitant esophageal atresia (EA) and tracheo-esophageal fistula (TEF) is a very rare condition, with a high mortality rate. Prematurity and congenital heart anomalies additionally increase the mortality rate. This situation is a great challenge for clinicians, requiring multidisciplinary work and adequate treatment strategy. **Case report.** We presented a premature twin newborn at the gestational age of 33/34 weeks with body mass of 1690 g. The existence of the left CDH was established on prenatal ultrasound exam in the 24th gestational week, and the diagnosis of EA with TEF was made on admittance to our hospital. The cardiac ultrasound exam revealed the total anomalous pulmonary venous connection (TAPVC). The first operation was performed on the day of admittance and consisted of left subcostal laparotomy, diaphragmatic repair, elastic occlusion of the gastroesophageal junction and gastrostomy. The ligation of TEF and esophagoplasty were done 13 days later in the second operation. The lethal outcome during the esophagoplasty was caused by the crisis of pulmonary hypertension and associated congenital heart anomaly (TAPVC). The presence of CDH and EA/TEF in association with TAPVC in a twin newborn has not been reported before in the literature. **Conclusion.** The treatment of newborns with CDH and EA/TEF requires multidisciplinary well-coordinated team work of pediatric surgeons, anaesthesiologists, neonatologists and pulmologists. The standard protocol for the management does not exist, but well-planned staged operations could enable greater survival rate.

Key words:
infant, premature; congenital abnormalities; hernia, diaphragmatic; esophageal atresia; tracheoesophageal fistula; heart defects, congenital; digestive system surgical procedures.

Apstrakt

Uvod. Kongenitalna dijafragmalna kila (KDK) udružena sa atrezijom jednjaka (EA) i traheo-ezofagealnom fistulom (TEF) veoma je retko stanje, sa visokom stopom smrtnosti. Prematuritet i urođene srčane mane dodatno povećavaju stopu smrtnosti. Ovo stanje predstavlja veliki izazov za kliničare i zahteva multidisciplinarni rad i adekvatnu strategiju lečenja. **Prikaz bolesnika.** U radu je prikazano prevremeno rođeno novorođenče iz blizanačke trudnoće, rođeno u 33/34 gestacionoj nedelji, sa telesnom masom od 1 690 g. Postojanje leve dijafragmalne kile utvrđeno je na prenatalnom ultrazvučnom pregledu u 24. gestacionoj nedelji, dok je dijagnoza EA sa TEF postavljena po prijemu u našu bolnicu. Ultrazvučnim pregledom srca dijagnostikovano je postojanje totalnog anomalnog utoka plućnih vena (*total anomalous pulmonary venous connection* – TAPVC). Prva operacija učinjena je na dan prijema i obuhvatala je levu supkostalnu laparotomiju, rekonstrukciju dijafragme, elastično zatvaranje gastroezofagealnog spoja i gastrostomiju. Podvezivanje TEF i ezofagoplastika su učinjeni 13 dana kasnije, tokom druge operacije. Smrtni ishod tokom ezofagoplastike je bio uzrokovan krizom plućne hipertenzije i udruženom srčanom manom (TAPVC). Postojanje KDK i EA/TEF kod novorođenčeta iz blizanačke trudnoće i udruženost ovog stanja sa TAPVC do sada nisu objavljeni u literaturi. **Zaključak.**

Correspondence to: Djordje Savić, Majke Jevrosime 10/17, 11 000 Belgrade, Serbia. E-mail: savic.60.djordje@gmail.com

Lečenje novorođenčadi sa CDH i EA/TEF zahteva multidisciplinarni, dobro koordinisan timski rad dečjih hirurga, anesteziologa, neonatologa i pulmologa. Standardni protokol lečenja ne postoji, ali dobro planirane etapne operacije mogle bi da omoguće veću stopu preživljavanja.

Ključne reči:
novorođenče, prevremeno; anomalije; hernija, dijafragmalna; jednjak, atrezija; traheoezofagusna fistula; srce, kongenitalne mane; hirurgija digestivnog sistema, procedure.

Introduction

Congenital Bochdalek diaphragmatic hernia (CDH) with concomitant esophageal atresia (EA) and tracheoesophageal fistula (TEF) is a very rare entity, with extremely high mortality rate. Prematurity and congenital heart anomalies are additional conditions that increase the mortality rate. In 2004 California Birth Defects Monitoring Program, analyzing the population of 3,318.966 live births and stillbirths in the period 1983–1996, there were reported 433 cases with Bochdalek type CDH (1.3:10000 births), 893 cases with EA/TEF (2.7:10000 births), and 18 cases with CDH and EA/TEF (0.05:10000 births). The mortality rate of patients with CDH associated with EA/TEF is very high, and it was reported that 16 of 17 babies were stillborn or died soon after birth [1]. Until 2015 there were only two database publications in relevant literature, announced in 2005 and 2013, and each reports approximately 20 cases of CDH with EA from different registries [1, 2]. Only 13 cases were reported in details [2–11], and all cases were sporadic, presented as case reports. Of those 13, there were only 3 case reports with successful management and survival of an infant with left CDH, EA, and TEF [3–5]. The first single-institution series of 6 newborns with CDH, EA and TEF appeared in 2015. Five newborns were operated on, and 3 patients survived 12.

The cause of the association of CDH and EA is still unknown. Chromosomal abnormalities were reported, such as a mosaic duplication on the long arm of the chromosome 10, but the genetic cause was not identified yet [1, 5]. The reasons of the high mortality rate are pulmonary hypertension and lung hypoplasia, caused by diaphragmatic hernia, combined with gastric acid reflux into respiratory tract and gastrointestinal distension in the chest cavity, caused by TEF. Associated cardiac anomalies, such as truncus arteriosus communis (TAC) [6] and total anomalous pulmonary venous (TAPVC) in our patient, and prematurity increased further the mortality rate. The presence of EA and TEF, causing gastric acid reflux and gastrointestinal distension in the chest cavity, exclude the usually applied initial conservative treatment of CDH, based on the delay of operative treatment until pulmonary hypertension decrease. CDH with EA/TEF is a great challenge for clinicians, requiring multidisciplinary work and adequate surgical strategy.

Case report

A premature twin male infant was born in a provincial hospital by spontaneous vaginal delivery, at the gestational age of 33/34 weeks, with body weight of 1,690 g. The existence of the left diaphragmatic hernia (Bochdalek type) was diagnosed on the prenatal ultrasound exam in the 24th gestational week. No other anomalies were found. The Apgar score at birth was 1 and 2 at the 1st and 5th minute, respectively. The newborn was intubated immediately after birth, but a nasogastric tube could not be placed. Chest radiography confirmed the left sided diaphragmatic hernia. His twin brother weighed 1,900 g, with no diagnosed anomalies, and was further treated in the hospital where he was born. The newborn with diagnosed CDH and suspected EA was transferred to our hospital 6 hours after birth. At the admission, the baby was intubated, cyanotic, without nasogastric tube. He was reintubated nasotracheally and pressure-controlled mechanical ventilation was started. The attempt of placing a nasogastric tube was unsuccessful. In the neonatal intensive care unit (NICU), a small amount of barium contrast was given in the nasoesophageal tube, and the radiography confirmed the clinical suspicion of EA. The presence of air in the stomach and intestinal loops situated in the chest pointed to the presence of TEF and left sided CDH (Figure 1). The abdominal ultrasound exam showed the presence of intestinal loops in the left thoracic cavity, without anomalies on abdominal parenchimatous organs. The cardiac ultrasound exam diagnosed the patent ductus arteriosus (DAP) and pulmonary artery hypertension (HAP), and raised the suspicion of the TAPVC.

The decision for urgent operative treatment was made, and 4 hours after the admittance and the use of resuscitation measures, the baby was transferred to the operating room. Through the left subcostal incision the left hemidiaphragm was approached. There was no hernial sac. Herniated organs (stomach, small intestine, colon and spleen) were pulled out of the thoracic cavity into the operative field. The anterior rim of the left diaphragm was well-developed, and the posterior rim was present as underdeveloped. The chest tube was placed, and the primary reconstruction of the left hemidiaphragm was made by mattress sutures. Malrotation was managed by Ladd operation. Then, a rubber sling was placed around the gastroesophageal junction, pulled through the side hole of the shortened nasogastric tube, and both exteriorized through the abdominal wall, in order to perform moderate angulation and temporary occlusion of the esophagus, so preventing the disastrous effect of TEF. A classical Stamm gastrostomy was made, and the underwater testing of gastrostomy Pezzer tube showed no air leakage through the TEF in the stomach (Figure 2).

Through a period of 13 days, the mechanical ventilation and all other intensive care measures were applied, but pulmonary hypertension crisis were repeated and blood oxygenation rate varied from 32% to 82%. The chest radiography showed the recovery of the left lung. Meropenem and vankomycin were administered initially, but *Acinetobacter* was diagnosed in the tracheal aspirate, so colistimethate sodium was added [13]. Haemocultures were sterile.

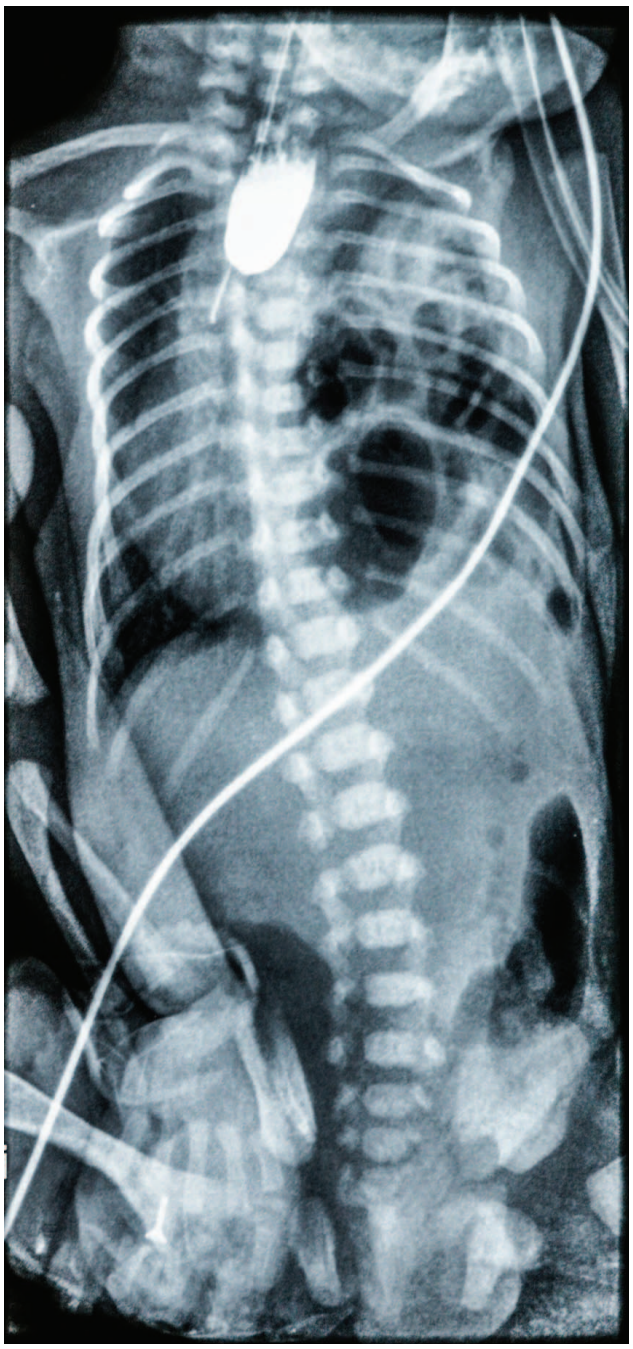

Fig. 1 – The presence of air in the stomach and intestinal loops situated in the chest pointed to the presence of the tracheo-esophageal fistula and left congenital diaphragmatic hernia.

Total parenteral nutrition was administered. Nonobstructive TAPVC was confirmed on the postoperative control cardiac ultrasound exam. The second operation was undertaken on the 13th postoperative day. Through the right posterolateral thoracotomy the TEF was approached and the large fistula was ligated and transected. The operation was continued with improved blood oxygenation rate of 80% and the esophagoplasty was started. During this procedure bradicardia, desaturation crisis, arterial hypotension and pulmonary oedema developed and, in spite of all resuscitation measures, led to the lethal outcome.

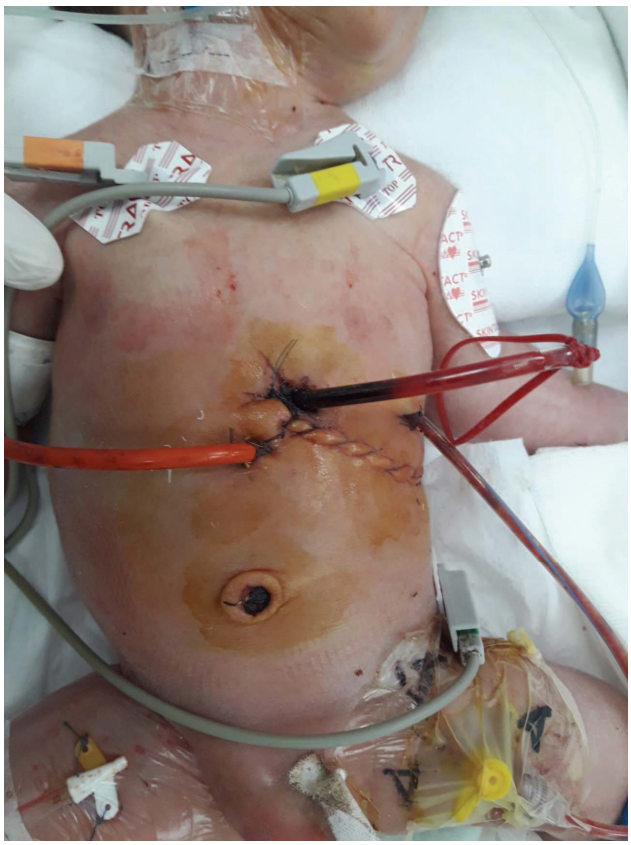

Fig. 2 – Stamm gastrostomy tested underwater showed that there was no air leakage through the Pezzer tube to the stomach.

Discussion

In babies with CDH and EA/TEF, the operative strategy is of the greatest importance. Sapin et al. [3] in 1996 reported the first successfully treated infant with CDH and EA/TEF, suggesting the laparotomic repair of CDH and avoiding thoracotomy in the first operation. The ligation of the TEF and esophagoplasty were postponed for the second operation. This attitude was based on the fact that laparotomy is better tolerated than thoracotomy, especially in the presence of pulmonary hypertension and left lung hypoplasia, which are the major problems in newborns with CDH. The disastrous effects of gastric acid reflux into respiratory system through TEF the author precluded by temporary occlusion, i.e., angulation of distal esophagus, placing a rubber sling around gastroesophageal junction. The gastrostomy was placed for gastric decompression. The ligation of TEF and esophagoplasty were performed after the baby's recovery, 2 weeks after the first operation [3]. This attitude was applied in the case reported by Charles et al. [5] in 2014, also with positive outcome and the baby's survival. An alternative strategy, the ligation of TEF and esophagoplasty in the first operation, and the repair of CDH in the second operation, with good outcome, was reported in 2013 [4]. The third operative strategy, reported by Zahn et al. [12] in 2015, suggested the CDH repair and ligation of distal esophagus 2 cm below TEF via transabdominal approach, and esophagoplasty 4–6 weeks later. This strategy was applied in 5 newborns and 3 patients survived.

In our patient with CDH, EA and TEF, we applied the principles of staged operative repair proposed by Sapin et l. [3] and Charles et al. [5]. The first operation, consisted of diaphragmatic repair, temporary occlusion of distal esophagus by silicone rubber sling and gastrostomy were performed without major problems in baby's respiratory and circulatory status, during and after the operation. So we could conclude that the proposed strategy was appropriate for babies with CDH and EA/TEF. The rubber sling around the gastroesophageal junction proved to be efficient in the temporary occlusion of distal esophagus and prevention of harmful effects of TEF. In our patient, there were no problems neither in the left hemidiaphragm repair nor in the closure of the abdominal wall, so that prosthetic material need not to be applied. The recovery of the left lung, as proved on the control postoperative chest radiography, was very satisfactory. The timing of the second operation, planned for the ligation of TEF and esophagoplasty, was determined according to literature suggestions and the baby's overall condition, and it was performed the 13th day of life. The sudden worsening of baby's condition and lethal outcome during the esophagoplasty was caused by the pulmonary hypertension crisis and associated congenital heart anomaly (TAPVC). The association of CDH with EA/TEF and TAPVC has not been reported yet in the literature. Also, the presence of CDH and associated EA/TEF in one of the twins was not mentioned in any of the references.

Conclusion

CDH (Bochdalek type) with associated EA and TEF is a very rare condition. Pulmonary hypertension and lung hypoplasia, caused by diaphragmatic hernia and gastric acid reflux into respiratory tract and gastrointestinal distension in the chest cavity, caused by TEF, lead to very high mortality rate. The treatment of these babies requires multidisciplinary, well-coordinated team work of pediatric surgeons, anaesthesiologists, neonatologists and pulmologists. The standard protocol for the management does not exist, but well-planned staged operations could improve survival rate.

R E F E R E N C E S

1. *van Dooren M, Tibboel D, Torfs C.* The co-occurrence of congenital diaphragmatic hernia, esophageal atresia / tracheoesophageal fistula, and lung hypoplasia. Birth Defects Res Part A Clin Mol Teratol 2005; 73(1): 53–7.
2. *Ben-Ishay O, Johnson VM, Wilson JM, Buchmiller TL.* Congenital diaphragmatic hernia associated with esophageal atresia: Incidence, outcomes, and determinants of mortality. J Am Coll Surg 2013; 216(1): 90–95.e2
3. *Sapin E, Berg A, Raynaud P, Lapeyre G, Seringe R, Helardot PG.* Coexisting left congenital diaphragmatic hernia and esophageal atresia with tracheoesophageal fistula: Successful management in a premature neonate. J Pediatr Surg 1996; 31(7): 989–91.
4. *Abdul Haium AA, Sim SW, Ong LY, Rajadurai VS.* Congenital diaphragmatic hernia associated with oesophageal atresia and trachea-oesophageal fistula in a low birth weight infant. BMJ Case Rep 2013; 2013. pii: bcr2013200014.
5. *Charles EJ, Judge JM, Vergales BD, Randall AH, Kane BJ, McGahren ED,* et al. Managing concomitant congenital diaphragmatic hernia, esophageal atresia, and tracheoesophageal fistula: A case report of a premature infant that achieved survival. J Pediatr Surg Case Reports 2014; 2(5): 239–42.
6. *Cunát V, Stranák Z, Pýcha K, Tláskal T, Melichar J, Miletín J,* et al. Congenital diaphragmatic hernia associated with esophageal atresia, tracheoesophageal fistula, and truncus arteriosus in a premature newborn. Pediatr Surg Int 2005; 21(8): 684–6.
7. *Takehara H, Komi N, Okada A, Nishi M, Masamune K.* Left diaphragmatic hernia associated with lower esophageal atresia. Pediatr Surg Int 1993; 8(4): 339–40.
8. *Ahmed S.* Right-sided Bochdalek hernia associated with esophageal atresia and trachea-esophageal fistula. J Pediatr Surg 1970; 5(2): 256.
9. *Udassin R, Zamir O, Peleg O, Lernau O.* Coexisting left diaphragmatic hernia and esophageal atresia. Pediatr Surg Int 1987; 2: 301–3.
10. *Gibon Y, Borde J, Mitrofanoff P, Lefort J.* Association of left diaphragmatic hernia, lung agenesia and esophageal atresia. Chir Pediatr 1978; 19(4): 261–7. (French)
11. *Al-Salem AH, Alkhuwaher H.* Coexisting congenital diaphragmatic hernia, esophageal atresia, and tracheoesophageal fistula: A case report and review of the literature. Int Surg 2008; 93(3): 141–4.
12. *Zahn KB, Scherf S, Schaible T, Wessel LM, Hagl CI.* Single-staged surgical approach in congenital diaphragmatic hernia associated with esophageal atresia. J Pediatr Surg 2015; 50(8): 1418–24.
13. *Đurđević Mirković TD, Gvozdenović L, Majstorović-Strazmester G, Knezević V, Celić D, Mirković S,* et al. An experience with colistin applied in treatment of imunocompromised patients with peritonitis on peritoneal dialysis. Vojnosanit Pregl 2015; 72(4): 379–82.

Received on March 22, 2017.
Accepted on September 25, 2017.
Online First September, 2017.