CASE REPORT

Congenital diaphragmatic hernia associated with oesophageal atresia and tracheo-oesophageal fistula in a low birth weight infant

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SUMMARY

The cooccurrence of congenital diaphragmatic hernia and oesophageal atresia with distal tracheo-oesophageal fistula is very rare and carries high mortality. Very few anecdotal case reports and one case series have been reported in the literature. We report a case of a late preterm, low birth weight infant with this rare association who was successfully managed by staged surgical approach and had good outcome.

BACKGROUND

The incidence of congenital diaphragmatic hernia (CDH) ranges between 1:2000 and 1:5000 live births.¹ The mortality in neonates with CDH remains high with overall survival rate around 60% and individual centre variation of 25–83%.^{1 2} Although the severity of pulmonary hypoplasia and pulmonary hypertension are the major determinants of overall survival, infants with CDH with another major defect have a greater morbidity and mortality.¹ Documented associated abnormalities of the oesophagus in infants with CDH include gastro-oesophageal reflux disease (GORD), oesophageal motility disorders, oesophageal duplications cysts and oesophageal atresia with tracheo-oesophageal fistula (OA/TOF). In particular, the combination of OA/TOF in the presence of CDH is extremely rare. This presents a difficult clinical problem to manage and carries high mortality. We report a case of a premature, low birth weight baby with the above combination who was managed with staged surgical approach and had a good outcome.

CASE PRESENTATION

A new-born male infant weighing 1855 g with antenatal diagnosis of CDH since the 20-week gestation period was born by vaginal delivery at the 36th week of gestation after spontaneous onset of preterm labour. The pregnancy was complicated by polyhydramnios, requiring amnio-reduction at 31 weeks. The amniocentesis showed normal male karyotype. The baby had mild respiratory distress at birth with apgar scores of 8 at 1 min and 9 at 5 min, respectively. He was intubated in the delivery room, mechanical ventilation started and stabilised initially on conventional ventilation.

Repeated oro-gastric tube insertion in the neonatal intensive care unit was unsuccessful. Diagnosis of OA/TOF was made postnatally, based on clinical and chest X-ray findings (figure 1) which were later confirmed at surgery. He had bilateral undescended

testes. The eye examination including fundus examination was normal. The anus was patent. There were no other external congenital malformations.

INVESTIGATIONS

Investigations were undertaken to exclude any other associated anomalies. The chest and abdominal X-ray showed no vertebral or bony abnormalities. The ultrasound scan of the abdomen showed normal kidneys. There were no structural cardiac defects noted on the echocardiography. The otoacoustic emission and automated auditory brainstem response hearing screening tests were normal. There was no evidence of VACTERL association or CHARGE syndrome.

TREATMENT

Following a multidisciplinary discussion, the decision was made to do a staged approach with ligation of TOF and repair of OA first followed by the delayed repair of the CDH. Ligation of TOF and repair of OA was performed at 16 h of age via

Figure 1 Image showing left-sided diaphragmatic hernia with bowel gas pattern in the left side of the chest. There is mediastinal shift to the right. Endotracheal tube, umbilical arterial and umbilical venous catheters are in place. A Replogle suction catheter is present in the blind upper pouch of oesophagus at the T4 vertebral body level.

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open right thoracotomy and trans-pleural approach. A rigid bronchoscopy prior to surgery confirmed a single large fistula just proximal to carina. Intra-operative findings were that of a type III OA with proximal pouch at the level of the T3 vertebral body. There was some distortion of the thoracic anatomy with the mediastinal structures rotated anteriorly and to the left. Meconium-filled bowel loops could be visualised posterior to the distal oesophagus. The fistula was found just proximal to the carina. There was a gap of 0.5 vertebral bodies between the two oesophageal ends after full mobilisation. The anastomosis was performed with interrupted 6-0 polydioxanone (PDS) sutures over a size 6 French feeding tube. The chest was closed after insertion of a size 10 French chest tube.

Postrepair, the baby was kept on mechanical ventilation and on muscle relaxants for 3 days. He had increased ventilatory requirements associated with respiratory secretions and needed high-frequency oscillatory ventilation support post-OA repair. He did not require inhaled nitric oxide. He also had systemic hypotension requiring dopamine, dobutamine and epinephrine in the first week of existence.

Repair of the CDH was carried out at 7 days of age via a left subcostal incision. A large left-sided postero-lateral diaphragmatic defect of 4 cm×5 cm was found in the stomach, with the majority of the small bowel, colon, appendix and spleen herniating to the chest. There was no sac and the left lung was fairly well developed. No definite posterior rim of diaphragm was identifiable. The diaphragmatic defect was closed with Gore Dualmesh biomaterial using 3-0 Ethibond suture. This was anchored to the remnant diaphragm anterior and laterally, and postero-laterally directly to the chest wall.

The immediate postoperative period was complicated by pulmonary collapse which improved with ventilatory management. He developed left chylothorax on the second postoperative day. This resolved completely with needle aspiration and treatment with octreotide. He was extubated to continuous positive airway pressure (CPAP) support on the seventh postoperative day (14 days of age) and weaned off CPAP support to room air at 24 days of age. Enteral feeds were started on day 15 after confirming the absence of anastomotic leak on contrast study of the oesophagus. Full enteral feeds were reached by 21 days of age.

OUTCOME AND FOLLOW-UP

He needed prolonged hospitalisation with feed intolerance and gastro-oesophageal reflux (GOR) requiring medication, continuous nasogastric tube feeding and a short period of naso-jejunal feeds. He was discharged home at 4 months of age on bolus nasogastric tube feeding. On follow-up at 10 months of age his GOR had improved. He was orally feeding well and thriving. Both of his testes have descended into the scrotal sac. His development was appropriate for age.

DISCUSSION

CDH with OA is a rare combination and each can be considered a major associated anomaly for the other. The incidence of anomalies associated with OA is 48% and that of CDH is 21%, respectively.³ The coexistence of both conditions is rare and may represent early embryologic insult, the timing of budding of the endoderm of the foregut into the trachea and the oesophagus and the formation of the diaphragm. There are some genetic and environmental hypothesis that has been proposed for this association.⁴ Chromosomal anomalies such as trisomies 13, 18 and 21 and some other autosomal anomalies have been shown to be associated with OA/TOF.⁴ Animal studies revealed some genes which may affect both

diaphragmatic and oesophageal development, but no human studies of any putative genes for the above association are available.⁴ Trisomy 18 had been reported with coexisting CDH and OA/TOF.^{5 6} One case of CHARGE syndrome had also been reported with this association.⁶

An epidemiological study of combinations of these defects from 1983 to 1996 by the California birth defects monitoring programme identified 18 cases of CDH/lung hypoplasia (LH) with OA/TOF (5/1 million live births). Of 17 cases of CDH/LH with OA/TOF, 16 were either stillborn or died.⁴ In a more recent series of 4888 patients on CDH registry between January 2000 and August 2011, 23 (0.05%) patients with CDH had an associated OA, out of which 19 (83.3%) presented with a TOF. There were only 6 survivors out of 23 in this cohort with a survival rate of 26.1%.⁶

Apart from the above-mentioned case series of 23 patients from the CDH registry,⁶ review of medical literature revealed only about 20 other anecdotal cases of this condition; most of them reported as part of associated anomalies in series on CDH or OA and TOF. The detailed description and surgical details of the patients and outcome is available only in 11 patients with only 2 survivors.^{5 7-14}

The antenatal diagnosis of OA/TOF in the presence of CDH is difficult. In our institution, the majority of CDH are diagnosed antenatally on ultrasound screening. The antenatal suspicion of OA/TOF is not so clear-cut. This is usually suspected on the basis of polyhydramnios and a small stomach bubble. CDH is also associated with polyhydramnios and the anatomical distortion caused by bowel herniation into the chest may influence detection of a small stomach, thus making it difficult to separately identify both anomalies.

The surgical management of coexisting CDH and OA/TOF should be individualised. Factors to be considered in determining both operative approach and timing include the clinical stability of the neonate; the side of the diaphragmatic hernia, and the combined experience of the neonatal, surgical and anaesthetic teams.

The need for mechanical ventilation in the presence of the TOF with CDH results in distension of stomach and intestines with air. The OA prevents decompression of the gut via a nasogastric tube and this could result in significant mediastinal shift, further worsening the respiratory status of the infant and making preoperative stabilisation complicated. In view of this, the repair of the TOF/OA should be the priority. If the neonate is not stable enough for the procedure, then the ligation of the fistula can be performed first and gut continuity re-established at a later date.

In our case, we elected to do a staged approach. We carried out ligation of TOF via a right thoracotomy incision together with oesophageal anastomosis at the same setting. A delayed repair of left CDH via left subcostal laparotomy was performed only after the baby had recovered from the initial surgery and was haemodynamically stable. This decision is inline with our current management of a neonate with isolated CDH. We believe that early ligation of TOF helped in stabilising the respiratory status and further optimised the baby for CDH repair. Oesophageal anastomosis restored the continuity of the gut and allowed for the passage of a nasogastric tube to decompress the stomach.

In conclusion, the cooccurrence of CDH and OA with TOF is very rare and associated with high mortality. The treatment approach has to be individualised to the neonate and involve a multidisciplinary team. With optimal care, patients with reasonably good pulmonary status and no other major associated anomalies, complete repair of both anomalies can be achieved.

Learning points

- ▶ Coexisting congenital diaphragmatic hernia (CDH) and oesophageal atresia/tracheo-oesophageal fistula (OA/TOF) is very rare and has high mortality especially in low birth weight infants.
- ▶ The treatment approach has to be individualised to the neonate. Early ligation of TOF will help in stabilising the patient before CDH repair.
- ▶ With optimal care, in the case of patients with reasonably good pulmonary status and no other major associated anomalies, complete repair of both CDH and OA/TOF can be achieved.

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Contributors All authors were involved with the clinical management of the patient. AAAH and SSWS jointly wrote the manuscript and LYO and VSR have reviewed the manuscript.

Competing interests None.

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