CASE REPORT

Rare combination of left-sided congenital diaphragmatic hernia and omphalocele

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SUMMARY

We reported a rare case of left-sided posterolateral congenital diaphragmatic hernia (CDH) and omphalocele, which is not associated with chromosomal abnormalities or other syndromes. Omphalocele was detected antenatally (CDH was not detected in antenatal ultrasound). The patient suffered from respiratory failure secondary to severe pulmonary hypertension. As the combination of CDH and omphalocele is rare and with the abdominal content herniating into the omphalocele instead of the thorax, antenatal diagnosis of such condition can be difficult. Unlike other reported cases in the literature, our patient's respiratory condition has been improving with time and is surviving beyond the infancy period. We believe this to be the first such survival case reported in the literature.

BACKGROUND

The incidence of congenital diaphragmatic hernia (CDH) is approximately 3/10 000 births; omphalocele occurs at a similar frequency.^{1 2} The simultaneous occurrence of both anomalies would be expected to be extremely rare and such cases have been reported to have a poor prognosis. Antenatal diagnosis of both CDH and omphalocele can be difficult if all the abdominal contents were herniated into the omphalocele with no displacement into the thorax. It requires a high index of suspicion for diagnosis both antenatally and postnatally. This report describes a first case of left-sided posterolateral CDH and omphalocele which survives beyond the infancy period.

CASE PRESENTATION

A 38-year-old Chinese woman was referred to our hospital with an antenatal diagnosis

of a giant omphalocele (containing liver and stomach) at 21 weeks of gestation. Antenatal ultrasound showed a small fetal chest (objective measurement of the fetal lung size was not available) and normal amniotic fluid volume. This was her second pregnancy. Chorionic villus sampling revealed a normal male karyotype and a normal microarray. Prenatal history was otherwise uneventful. A 2.7 kg male was delivered at 37 weeks gestational age by emergency caesarean section (for breech presentation in labour). The baby was intubated for secondary apnoea at birth. His Apgar score was 4 at 1 min and 7 at 5 min. A giant omphalocele containing liver, spleen and small bowel was noted ([figure 1](#)). No other associated malformation was found. He subsequently developed pulmonary hypertension requiring high-frequency oscillatory ventilation and hypotension given multiple inotropes.

Construction of a silo was performed on the fifth day of life. Liver, spleen and small bowels were found outside his abdominal cavity. Primary reduction of the omphalocele contents was attempted (instead of allowing spontaneous epithelialisation with delayed closure to minimise the risk of infection of the omphalocele) but not possible as it resulted in immediate ventilatory difficulty and hypotension intraoperatively. Daily bedside reductions of the omphalocele along with revisions of the silo were carried out on days of life 16, 35, 68 and 77. His omphalocele underwent self-epithelialisation subsequently.

Serial chest radiographs revealed elevated left hemidiaphragm ([figure 2](#)). Ultrasound of the diaphragm on day of life 61 showed a reduction of movement of the left hemidiaphragm with left lobe of liver, left kidney and spleen being found in the left lower hemithorax ([figure 3](#)), consistent with a left-sided CDH. Repair of the diaphragmatic hernia was successfully performed on day of life 77, with bowels in the thoracic cavity being pushed back into the abdomen.

Mechanical ventilation with intratracheal intubation was continued until day 96 after birth; nasal non-invasive ventilation was given for chronic respiratory failure with moderate to severe pulmonary hypertension after extubation. Repeated intubations and mechanical ventilation were required afterwards due to aspiration or lung collapse with sputum retention. Bronchoscopy and CT of the thorax revealed tracheo-/bronchomalacia and left lung hypoplasia. Management of pulmonary hypertension and chronic lung disease including

Figure 1 Omphalocele containing liver, spleen and small bowel.

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Figure 2 Chest radiograph showing elevated left hemidiaphragm.

sildenafil, diuretics and inhale steroid were given. Respiratory condition gradually improved.

OUTCOME AND FOLLOW-UP

The patient was able to be weaned off from non-invasive ventilation intermittently to nasal cannula since 11 months of life.

Full feeding was achieved and total parenteral nutrition (TPN) was given for approximately 4 months. Gastro-oesophageal reflux was treated with proton pump inhibitor. There was transient cholestasis and liver function derangement related to prolonged TPN, which resolved with time. There is good catch up in body weight with normal head growth.

DISCUSSION

Coexistence of CDH and an abdominal wall defect is considered to be rare and our case is the first reported survival case of an omphalocele associated with a left-sided posterolateral CDH up to date. Harmath *et al*, Sweed and Puri reported that

around 3–4% of CDH were found to have associated abdominal wall defect with an omphalocele (4 out of 100 and 4 out of 116 CDH cases, respectively^{3,4}). Borys and Taxy performed a 10-year review of autopsy cases and reported that the incidence of CDH associated with an omphalocele was 0.077%.⁵ With paucity of literature review on their perinatal management, this rare combination of CDH and omphalocele poses challenges in its clinical management.

In patients with combination of CDH and omphalocele, the severity of pulmonary hypoplasia/hypertension along with the presence of associated syndromal/chromosomal anomalies are believed to be the major determinants of outcomes in these infants. The combination of CDH and omphalocele had been reported as a component of syndromes, such as Fryns syndrome or pentalogy of Cantrell.⁶ Chromosomal abnormalities such as trisomy 13⁷ and trisomy 18³ had been reported as well. However, our patient presented here belongs to the non-syndromal group. While the non-syndromal coexistence of an omphalocele and CDH had been reported (Nishimura *et al*⁸, Scahill *et al*⁹, Nonaka *et al*¹⁰), all these cases died within the neonatal period either as a result of the associated cardiac anomaly (hypoplastic left heart syndrome) or severe pulmonary hypertension and respiratory failure. Our case is surviving beyond the infancy period and the absence of major structural heart disease offers survival benefit.

Accurate diagnosis of both CDH and omphalocele in the antenatal period can be difficult if all the abdominal contents are herniated into the omphalocele with no displacement into the thorax.¹⁰ Similar to our index case, CDH was only diagnosed after birth in two other cases reported in the literature (Nishimura *et al*⁸, Scahill *et al*⁹). Timing of surgical treatment in cases with both CDH and omphalocele is important. There was report on CDH causing severe respiratory distress or circulatory instability after closure of the abdominal wall defect.⁸ In our index case, primary closure of the omphalocele was not possible and ultimately it underwent self-epithelialisation.

Contributors YYC is responsible for care of the patient in the intensive care unit, participated in writing the article and received informed consent from the guardian. SCMW and MSRW are responsible for care of the patient in the ICU and critically revised the article.

Competing interests None declared.

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Figure 3 Ultrasound of diaphragm showing left lobe of liver and left kidney in left lower hemithorax.

Learning points

- Combination of congenital diaphragmatic hernia (CDH) and omphalocele is rare and antenatal diagnosis may be difficult. It requires a high index of suspicion to diagnose this condition postnatally.
- Survival is possible in patient with both CDH and omphalocele. The absence of associated syndromal/chromosomal anomalies or major structural heart disease offers survival benefit.
- The presence of pulmonary hypoplasia/hypertension results in significant respiratory failure and it is the focus of the clinical management in such cases in the long term.

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