CASE REPORT

Accessory diaphragm associated with non-immune hydrops fetalis

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SUMMARY

An accessory diaphragm, also known as diaphragmatic duplication, is a congenital anomaly in which there is a fibromuscular membrane on top of the normally formed diaphragm dividing the hemithorax into two compartments trapping part of the pulmonary parenchyma. It is a very rare anomaly with less than 40 cases reported in the literature, of which only five were diagnosed in the newborn period, with no reports on any clinical clues for the antenatal diagnosis of this condition. We describe a case of congenital accessory diaphragm presenting in the antenatal period as hydrops fetalis. We also describe the radiological features of this rare anomaly on the antenatal fetal ultrasound.

BACKGROUND

Hydrops fetalis is a condition where excess fluid accumulates in the fetus with resultant high mortality and morbidity. Antenatal diagnosis is made by the ultrasonographic finding of fluid in at least one body cavity (ascites, pleural effusion or pericardial effusion) accompanied by skin oedema (greater than 5 mm thickness), or in two potential spaces without accompanying skin oedema. The possible aetiologies of hydrops fetalis are continually expanding as formerly unknown causes are being reported.¹

This interesting neonate presented in the antenatal period with hydrops fetalis. No definite cause of hydrops could be identified until postmortem examination which revealed the presence of an accessory diaphragm. This is a very rare congenital anomaly with only five cases reported in neonates. Fetal hydrops fetalis as a presentation of an accessory diaphragm has never been reported. In our case, antenatal ultrasound showed a distinct septum-like lesion in the right thoracic cavity of the fetus, which in retrospect was the accessory diaphragm. Early recognition of this congenital malformation on antenatal ultrasound will be helpful to direct an appropriate counselling and management plan.

CASE PRESENTATION

A 34-year-old primigravida woman was referred to the fetal-maternal unit of our tertiary care hospital for review. Antenatal ultrasound at 27 weeks gestation detected significant fetal skin oedema more than 5 mm thick, bilateral pleural effusions and ascites diagnostic of hydrops fetalis. A repeat scan at 33 weeks showed worsening pleural effusions and a membrane-like structure in the right pleural cavity (figure 1). The nature and significance of the latter was unclear.

The patient underwent lower segment caesarean section (LSCS) at 36 weeks gestation following the administration of two doses of corticosteroids for gradual worsening of hydrops fetalis. Immediately prior to LSCS, ultrasound-guided needle aspiration with removal of 60 mL of pleural fluid from each hemithorax was performed to assist with resuscitation at birth. A male baby with a birth weight of 2.7 kg (50th centile) was delivered. APGARs were 8 and 8 at 1 and 5 min, respectively. The infant looked hydropic with generalised pitting oedema of the skin. The umbilical cord gases were normal.

The infant developed significant work of breathing soon after birth requiring immediate intubation and mechanical ventilation. Chest X-ray at birth showed a large, right-sided pleural effusion, along with bilateral small volume lungs.

Cardiac echocardiography revealed significant pulmonary hypertension. This was managed with inhaled nitric oxide therapy and high-frequency oscillatory ventilation within a few hours after birth. The infant subsequently developed severe systemic hypotension requiring dopamine and epinephrine infusions. In spite of this, respiratory function continued to deteriorate with persistent severe respiratory acidosis and refractory pulmonary hypertension. In view of his deteriorating clinical condition, care was withdrawn at around 36 h of age.

The fetal pleural fluid aspirated just prior to birth revealed lymphocytes predominantly, suggesting an underlying diagnosis of chylothorax. There was no evidence of blood group incompatibility; maternal TORCH screening (toxoplasmosis, rubella, cytomegalovirus and hepatitis) was negative. Parvovirus serology was similarly negative.

The final clinical diagnosis at the time of death was non-immune hydrops fetalis secondary to chylothorax and associated pulmonary hypoplasia.

A three-cavity postmortem examination revealed an abnormality centred in the right hemithorax in the form of an additional membrane separating the right pleural cavity into two compartments (figures 2 and 3).

The membrane extended from the right chest wall peripherally to the right lung, dividing the right lung into two 'lobes' (figure 2). The membrane was adherent to the lower aspect of the right upper 'lobe'. An anatomically normal diaphragm was present below both lungs, where it normally separated the chest and abdominal cavities.

The right pulmonary vasculature appeared normal; however, the right lung was significantly underdeveloped (figure 3). At the time of the autopsy, the membrane was noted to be floppy,

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Figure 1 Fetal ultrasound, lateral view, showing the septum-like lesion (A) in the right hemithorax adherent to the small volume right lung (B) with fluid collection on either side of the septum.

presumably as a result of antenatal drainage. The enlarged right pleural cavity was seen to extend behind mediastinal structures into the left hemithorax. This extension had been misinterpreted radiologically as a left-sided pleural effusion, giving the impression of bilateral effusions. The left pleural cavity was otherwise normal.

These findings were consistent with the diagnosis of an accessory diaphragm and significant right lung hypoplasia.

DISCUSSION

An accessory diaphragm is a rare congenital anomaly. Neonatal presentation of this condition is even rarer. To the best of our

Figure 2 Right hemithorax organs as seen on autopsy, viewed from behind (A) accessory diaphragm, (B) right 'upper' lobe, (C) right 'lower' lobe, (D) true diaphragm. Note the extension of the voluminous cavity inferiorly into the costodiaphragmatic recess and (not shown) behind the mediastinum into the left hemithorax.

knowledge, only five neonates with accessory diaphragm have been reported until now, of which four presented with respiratory distress. Of these five cases, three were deceased and diagnosed only at autopsy, one had an unknown outcome and only one neonate had a diagnosis confirmed on MRI with subsequent surgical intervention and good outcome.²

Our case is unique in two ways. First, hydrops fetalis as a presentation of this condition has never been reported. Second, antenatal ultrasound provided a clue to this diagnosis showing a membrane-like structure in the right thoracic cavity. Unfortunately, we were unable to determine the significance of this finding at the time of scanning. Retrospective correlation with postmortem findings indicate that the membrane-like structure noted antenatally was in fact an accessory diaphragm.

The exact cause of the pleural effusion in our case is unclear. It is most likely a direct result of the diaphragmatic malformation, possibly acting as a space occupying an intrathoracic mass. The presence of an intrathoracic mass, such as cystic adenomatoid malformation or mediastinal teratoma, is a well-known cause of hydrops with a well-established pathophysiology. The intrathoracic space occupying lesion increases the intrathoracic pressure leading to obstruction of venous and lymphatic flow and subsequent accumulation of pleural fluid and subsequent hydrops fetalis.³ The diagnosis of primary chylothorax, based on the high pleural fluid lymphocyte count, was clinically misleading as the autopsy detected the presence of an accessory diaphragm and not a primary vascular or lymphatic malformation.

An accessory diaphragm is a malformation more common in males, and most often right sided. It can occur in isolation or in association with lung hypoplasia or other vascular malformations.^{2,3} The greatest diagnostic challenge lies in the initial consideration of what is a very rare malformation, particularly in the antenatal period. Even in infancy, diagnosis based on routine X-rays is difficult and potentially misinterpreted as atelectasis or pulmonary hypoplasia.^{4,5} MRI is thought to be the optimal non-invasive diagnostic test for this condition postnatally.² Unfortunately, it was not possible to perform this test on our patient due to the infant's poor clinical condition.

The mechanism behind the formation of an accessory diaphragm is unclear. It has been postulated to be a defect in the synchronisation between the caudal migration of the septum transversum responsible for the formation of the diaphragm and the development of the bronchial system. Very early formation of the bronchial system may result in the developing septum

Figure 3 Hypoplastic right lung divided into two 'lobes' (B and C) by the accessory diaphragm (A). Note the larger, morphologically more normal left lung.

transversum being cleaved as it makes contact with the developing lung. The associated pulmonary vascular malformations reported in cases of accessory diaphragm in the literature suggest that the pathogenic event occurs during the same embryonic period.^{2–6} As best as could be ascertained, our infant did not have any associated vascular malformations.

Finally, we would like to highlight the benefits of aspiration of fetal pleural fluid prior to delivery. In our facility, it has been a standard practice to drain pleural fluid prior to delivery in cases of massive fetal pleural effusions. It has been shown that fetal centesis immediately prior to birth improves resuscitative efforts and ultimately survival.⁷

Learning points

- ▶ Non-immune hydrops fetalis is a clinical entity that carries serious morbidity and mortality.
- ▶ Ultrasound is the standard imaging tool used to determine the degree and extent of fetal hydrops and also to identify underlying fetal anomalies causing hydrops.
- ▶ An accessory diaphragm is a rare congenital anomaly that can be associated with fetal hydrops.
- ▶ High-resolution ultrasound can identify this anomaly in the antenatal period, which may be helpful to direct appropriate counselling and management in the antenatal and newborn periods such as further imaging including MRI.

Thus, an accessory diaphragm, although a rare congenital abnormality, should be considered in the differential diagnosis of a septum-like lesion in the pleural cavity on antenatal fetal ultrasound. MRI, either antenatally or after birth, may be used to confirm the diagnosis. Recognition of this condition is important since surgical treatment has resulted in uniformly excellent results.⁴ Our case demonstrates that it has a potential association with non-immune hydrops and should be considered in the differential diagnosis.

Contributors AP was involved in the case review, literature review and writing up of the manuscript. ES contributed to the diagnosis, literature review and writing up of the manuscript. DC contributed to the case diagnosis, management, literature review and writing up of the manuscript. SB was involved in the diagnosis, management, supervision and coordination of the write-up.

Competing interests None.

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