CASE REPORT

Strangulated diaphragmatic hernia presenting at 7 weeks of life as intractable shock

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SUMMARY

A 7-week-old infant presented to hospital pale and floppy, with 5 s capillary refill time. Blood gas showed severe acidosis (pH 6.86, partial pressure of carbondioxide 10.55 kPa, base excess 21.1). Hypotension persisted despite several fluid boluses so she was intubated and started on inotropic support. A chest X-ray revealed a congenital diaphragmatic hernia (CDH). Despite steroids and blood transfusions she remained unstable, and could not be resuscitated following cardiac arrest. Postmortem revealed 39 cm of herniated, necrotic colon. 5–25% of CDH presents after the neonatal period, and while not associated with pulmonary hypoplasia is primarily still a diaphragmatic defect. In late presenting cases, herniation occurs shortly prior to developing symptoms; therefore, an antenatal ultrasound (US) cannot pick it up. If we could diagnose the isolated diaphragmatic defect antenatally, this would allow elective postnatal surgical closure. This is not feasible currently; however, with the advent of antenatal three-dimensional US scans it may be possible in the future.

BACKGROUND

In general, late presenting congenital diaphragmatic hernia (CDH) has a better prognosis than neonatal presentation. However, it does pose a diagnostic challenge given the wide variety of clinical signs and symptoms these children have. Given the severity of presentation in our case, the diagnosis had little bearing on our management, but this case serves as a useful reminder to keep late-presenting CDH as a differential diagnosis when children present with severe respiratory or gastrointestinal (GI) symptoms, or indeed shock, especially when viewing radiographs since interventions based on misdiagnosis can have fatal consequences (eg, insertion of chest drains). Finally, we also discuss the hypothetical possibility of antenatally diagnosing the isolated diaphragmatic defect that allows herniation later in life.

CASE PRESENTATION

A 7-week-old infant presented to the accident and emergency department at 11pm, having become unwell since 7pm following an uncharacteristically prolonged breastfeed (45 min). She became unsettled, developed shallow breathing, looked pale, felt cold and was increasingly lethargic. She was previously well, born at term, with an uncomplicated pregnancy (antenatal anomaly scan was normal). She was exclusively breastfed, growing on the 50th centile, with no family history of note. On

presentation, she looked pale and floppy, with mild subcostal recession. Her central capillary refill time (CRT) was 5 s., temperature was 34°C, heart rate was 169 bpm and blood pressure (BP) was 42/15 mm Hg. Otherwise her chest sounded clear, her abdomen felt soft, with bowel sounds; there were no murmurs and femorals were palpable.

INVESTIGATIONS

Blood glucose was measured immediately at 16.6 mmol/L, and she was given a fluid bolus (20 mL/kg, 0.9% saline). Initial blood gas showed a severe mixed metabolic and respiratory acidosis (pH 6.86, partial pressure of carbon dioxide (PaCO₂) 10.55 kPa, base excess 21.1 and lactate 10.5 mmol/L). Her BP and central CRT had not improved so a further fluid bolus was given, along with intravenous amoxicillin and cefotaxime for presumed sepsis, and she was intubated. A repeat blood gas showed deterioration (pH 6.79, pCO₂ 14.60 kPa, base excess 18.4, lactate 10.87 mmol/L). In view of persistent hypotensive shock and severe acidemia, an adrenaline infusion was started (0.5 µg/kg/min), and she was given a sodium bicarbonate bolus.

DIFFERENTIAL DIAGNOSIS

At this point a chest X-ray was carried out, which revealed a left-sided diaphragmatic hernia (figure 1), and intravenous metronidazole was administered. This was followed by cardiac arrest, requiring one cycle of cardiopulmonary resuscitation and a 1 mL epinephrine bolus (1:10 000 concentration). The infusion was increased to 1g µg/kg/min, and norepinephrine was started at the same rate. Her blood gas showed worsening severe acidosis (pH 6.8, pCO₂ 4.13 kPa, base excess 29, lactate 14.6 mmol/L), she had so far received a total infusion of 100 mL/kg fluid within the space of 3 h.

TREATMENT

Blood test results revealed developing disseminated intravascular coagulation (DIC) (haemoglobin (Hb) 6.9 g/dL, International normalized ratio 1.9, activated partial thromboplastin time 2.7, platelets 242×10⁹/L), but were otherwise normal; C reactive protein was <1 mg/L. Blood was transfused at 15 mL/kg, during which she developed a widespread erythematous rash, which resolved following a 4 mg/kg bolus of intravenous hydrocortisone. The subsequent blood gas showed some improvement (pH 7.09, pCO₂ 3.82 kPa, base excess 21, lactate 13.79 mmol/L).

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Figure 1 Chest radiograph showing left-sided diaphragmatic hernia.

OUTCOME AND FOLLOW-UP

She was transferred to a paediatric intensive care unit, but her BP remained extremely difficult to maintain despite further fluid boluses and blood transfusions (Hb dropped to 2.9 g/dL). Echocardiography showed a normal heart structure. The following day she was unable to be resuscitated after cardiac arrest. Blood cultures returned negative at 48 h of incubation. The postmortem revealed a 2.5 cm defect in the left posterior diaphragm, through which 39 cm of the colon had herniated and become necrotic, suggestive of venous infarction. There was no evidence of pulmonary hypoplasia.

DISCUSSION

CDH (incidence of 1 in 2000–5000 live-births) results from a defect in diaphragmatic development during early pregnancy (8–10 weeks gestation), which allows herniation of abdominal viscera into the thoracic cavity, usually on the left side.^{1 2} The traditional view that this causes compression of the developing lung, resulting in pulmonary hypoplasia and hypertension, has been replaced by the ‘dual hit’ hypothesis, that there is also a degree of abnormal pulmonary development.³ CDH is associated with other organ defects in up to 33% of cases (eg, heart, neural tube, abdominal wall, craniofacial and urinary tract abnormalities).^{4–6}

Classically CDH presents at birth, but 5–25% cases of CDH do not present in the neonatal period,^{7 8} which can pose a diagnostic challenge due to non-specific GI and respiratory symptoms, which may be of acute or chronic onset (eg, respiratory distress, cough, vomiting and abdominal pain). A chest X-ray is the diagnostic modality of choice, but findings can be misinterpreted (eg, pneumothorax and pulmonary effusion), which can lead to delayed management, and iatrogenic complications (eg, chest drain placement).⁹

As with our case, late-presenting CDH generally lacks pulmonary hypoplasia and other-organ anomalies associated with neonatal CDH, leading to the hypothesis that it does not share the

same pathogenetic factors.¹⁰ However, in both, the primary defect is predominantly a diaphragmatic defect in the left posterolateral position, and lack of a sac indicates this occurs during embryological development. But whereas in neonatal CDH herniation of abdominal viscera occurs in utero or shortly after birth, in late-presenting cases it occurs shortly prior to developing symptoms, supported by diagnostic studies (including chest radiographs) reported as normal prior to presentation.^{10 11}

The nature of herniation accounts for the variety of clinical presentations associated with late-presentation; and in cases like ours, with an acute deterioration, this could be due to rapid herniation causing lung compression, mediastinal displacement and strangulation of the gut. In contrast, there are many anecdotal reports of children with undiagnosed repeated intermittent presentations of respiratory or GI symptoms—later found to have CDH—and which would be accounted for by recurring transient herniation through a relatively large diaphragmatic defect. The stimulus that causes herniation of abdominal viscera through the already present diaphragmatic defect is unknown, although likely associated with a rise in intra-abdominal pressure. In our case, could this have been the prolonged breastfeed? However, this may just as well have been due to the already herniating gut.

Antenatal ultrasound (US) scans reveal CDH in 50% of cases, by demonstrating herniation of abdominal viscera into the thoracic cavity, mediastinal shift and polyhydramnios.¹² Given lack of these features in late-presenting CDH, is there a way to antenatally diagnose the isolated diaphragmatic defect? This would allow elective postnatal surgical closure, and a diagnosis if the child presents unwell prior to this. Currently the diaphragm is visualised in the sagittal section as a line between the thoracic and abdominal cavity. Given the two-dimensional nature of routine scanning, it would be difficult to fully visualise the diaphragm with any accuracy due to its three-dimensional curvature. However with the advent of antenatal three-dimensional US scans (currently predominantly used in a non-diagnostic context in the UK, but showing increasing use in detection of foetal anomalies), there is a hypothetical possibility of diagnosing isolated diaphragmatic defects without associated herniation, especially given the majority occur through the foramen of Bochdalek.^{13–15}

Learning points

- ▶ Congenital diaphragmatic hernia (CDH) has an incidence of 1 in 2000–5000 live births worldwide, but only 50% are diagnosed antenatally, and in 5–25% of cases, presentation is not during the neonatal period.
- ▶ In any child that presents with respiratory or gastrointestinal symptoms, or circulatory collapse, undiagnosed CDH should be a differential diagnosis. Although rare, there is significant morbidity and mortality associated with late diagnosis and iatrogenic complications due to chest X-ray misinterpretation.
- ▶ Neonatal and late-presenting CDH may have a differing pathogenetic basis. The primary developmental defect of the diaphragm is similar, but the latter does not have pulmonary hypoplasia and other-organ defects associated with many cases of the former.
- ▶ Diagnosis of isolated diaphragmatic defects is not feasible with current antenatal two-dimensional ultrasound screening. However, it may be possible with greater use of three-dimensional scanning in the future, thereby allowing elective surgical closure postnatally.

Contributors All authors were involved in the clinical case, background literature search and writing of the manuscript.

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REFERENCES

- Butler N, Claireaux AE. Congenital diaphragmatic hernia as a cause of perinatal mortality. *Lancet* 1962;1:659–63.
- Lally KP. Congenital diaphragmatic hernia. *Curr Opin Pediatr* 2002;14:486–90.
- Keijzer R, Liu J, Deimling J, et al. Dual-hit hypothesis explains pulmonary hypoplasia in the nitrofen model of congenital diaphragmatic hernia. *Am J Pathol* 2000;156:1299–306.
- Enns GM, Cox VA, Goldstein RB, et al. Congenital diaphragmatic defects and associated syndromes, malformations, and chromosome anomalies: a retrospective study of 60 patients and literature review. *Am J Med Genet* 1998;79:215–25.
- Lin AE, Pober BR, Adatia I. Congenital diaphragmatic hernia and associated cardiovascular malformations: type, frequency, and impact on management. *Am J Med Genet C Semin Med Genet* 2007;145C:201–16.
- Sweed Y, Puri P. Congenital diaphragmatic hernia: influence of associated malformations on survival. *Arch Dis Child* 1993;69:68–70.
- Berman L, Stringer D, Ein SH, et al. The late-presenting pediatric Bochdalek hernia: a 20-year review. *J Pediatr Surg* 1988;23:735–9.
- Mei-Zahav M, Solomon M, Trachsel D, et al. Bochdalek diaphragmatic hernia: not only a neonatal disease. *Arch Dis Child* 2003;88:532–5.
- Baglaj M, Dorobisz U. Late-presenting congenital diaphragmatic hernia in children: a literature review. *Pediatr Radiol* 2005;35:478–88.
- Baglaj M. Late-presenting congenital diaphragmatic hernia in children: a clinical spectrum. *Pediatr Surg Int* 2004;20:658–69.
- Weber TR, Tracy T Jr, Bailey PV, et al. Congenital diaphragmatic hernia beyond infancy. *Am J Surg* 1991;162:643–6.
- Sinha CK, Islam S, Patel S, et al. Congenital diaphragmatic hernia: prognostic indices in the fetal endoluminal tracheal occlusion era. *J Pediatr Surg* 2009;44:312–16.
- Benacerraf BR, Shipp TD, Bromley B. How sonographic tomography will change the face of obstetric sonography: a pilot study. *J Ultrasound Med* 2005;24:371–8.
- Merz E, Welter C. 2D and 3D ultrasound in the evaluation of normal and abnormal fetal anatomy in the second and third trimesters in a level III center. *Ultraschall Med* 2005;26:9–16.
- Blaas HG, Eik-Nes SH. Sonographic development of the normal foetal thorax and abdomen across gestation. *Prenat Diagn* 2008;28:568–80.

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