CASE REPORT

Hepatic heterotopia in congenital diaphragmatic anomaly

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SUMMARY

An antenatally diagnosed case of a left congenital diaphragmatic hernia is reported. The diaphragmatic eventration hernia sac consisted of a sheet of ectopic liver tissue in continuity with a hypoplastic left lobe which formed the medial anterior and posterior walls of the hernia is presented. The operative management of this combination of defects has not previously been described in English literature. Embryological considerations, limitations of accurate preoperative diagnosis, technical challenge in the operative repair of the defect and need for drainage is discussed.

BACKGROUND

Congenital absence of left hemidiaphragm, the presence of a diaphragmatic eventration, atrophic left lobe of liver and heterotopia of the liver in the hernial sac are all very rare and unusual forms of anomalies. However, anecdotal cases are reported individually or in association with other malformations.^{1–6} We describe a case in which the left lobe of liver was the medial anteroposterior walls of the

defect and the eventrated diaphragmatic hernia sac was a sheet of hepatic tissue. Embryological hypotheses in the pathogenesis are considered and the surgical management is discussed.

CASE PRESENTATION

A female fetus, the product of in vitro fertilisation to consanguineous Afghan parents, was diagnosed at 28 weeks of gestation with a congenital left-sided diaphragmatic hernia. The karyotype was reported to be normal on amniocentesis. The mother was known to be a hypertensive and type II diabetic.

The baby was born by emergency lower segment caesarean section at 39 weeks of gestation following prolonged rupture of membranes. The mother had received steroids at 33 weeks of gestation. The baby's birth weight was 3.73 kg. The APGAR score was 6, 8 and 9. Curosurf was given at 3 and 90 min. The infant was admitted to the neonatal intensive care unit, requiring conventional ventilation followed by high frequency oscillation ventilation for 2 days. Dobutamine was prescribed for 2 days to maintain his systemic blood pressure.

Figure 1 Chest X-rays: Left congenital diaphragmatic hernia.

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Figure 2 Heterotopia of hepatic tissue in the hernial sac with central vein.

INVESTIGATIONS

A chest X-ray confirmed a congenital left-sided diaphragmatic hernia: the heart and mediastinum were displaced to right, and the nasogastric tube was observed to lie in the stomach within the left hemithorax (figure 1). Postnatal ECGs demonstrated a structurally normal heart and pulmonary hypertension.

DIFFERENTIAL DIAGNOSIS

Left congenital diaphragmatic anomalies—posterolateral hernia or diaphragmatic eventration was considered.

TREATMENT

A laparotomy was performed on day 3 following respiratory stabilisation. A left congenital posterolateral diaphragmatic hernia was found associated with an abnormally thick diaphragmatic eventration. This took the appearance of a cabbage leaf with large vein-like structures passing radially within it. A thin rim of diaphragmatic muscle was present laterally. Medially the margins of the eventration were continuous with an abnormally shaped, left lobe of the liver. The diaphragm and crura were

absent medially. The hernia contained stomach, small and large bowel, left and right lobes of the liver and spleen. These were reduced into the abdominal cavity. The sac was resected en bloc with most of the left lobe of the liver (figure 2). Intrathoracic examination revealed a very small hypoplastic left lung. The pericardium was present and intact without any defect. An intercostal drain was inserted. The hernia was repaired with absorbable sutures. The hernia was repaired medially by approximation of the remnants of left lobe of the liver and Toldt's fascia. The position of the duodeno-jejunal flexure was noted to be normal.

OUTCOME AND FOLLOW-UP

The baby was extubated on day 5 of life and the intercostal drain was removed on day 7. A total of 250 mL of clear fluid had been drained. The drain was removed after it had been clamped for 48 h, no fluid subsequently drained and the baby was well self-ventilating. On day 10 the baby developed acute respiratory distress requiring 80% oxygen on continuous positive airway pressure. An intercostal drain was reinserted as a large pleural effusion was demonstrated on ultrasound. Seventy milliliters of fluid from the left pleural effusion did not contain any significant blood, bile or protein. The intercostal drain was removed on day 13 when it did not drain any fluid after 48 h.

The baby was transferred to self-ventilating in air on full feeds to the local neonatal unit on day 15. At 7 years follow-up, she is well, asymptomatic and thriving well with normal chest X-ray.

The pathological examination of the excised tissue demonstrated a very thick dome-shaped diaphragmatic eventration measuring 7×5×5 cm dome-shaped structure continuous with the resected left lobe of liver (figure 2). Histology demonstrated the eventration to consist of a sheet of liver tissue including hepatocytes and portal tracts without any bile ducts (figure 3).

DISCUSSION

The association of congenital diaphragmatic hernia (CDH) and hepatic heterotopia has been reported in four previous cases. In each case the pericardium has been reported as absent or

Figure 3 Histological sections ×4 to ×10 magnification showing sheets of hepatic tissue in the diaphragmatic sac.

deficient. Hypotheses have been presented implicating the deficient pericardium in the pathogenesis of this condition.¹⁻⁴ Our case is unique as the pericardium was present and normal.

The development of the diaphragm is thought to involve two phases: (1) development of the diaphragmatic primordium and (2) the development of pleural cavity and the closure of the pleuroperitoneal canal involving the growth of the posthepatic mesenchymal plate.⁷⁻⁹ The phrenic nerve, or its progenitor cells, may mediate in the development of the diaphragm by a similar link to that of the genitofemoral nerve with testicular development.¹⁰ The septum transversum and the posthepatic mesenchymal plate are closely related and controlled by phrenic nerve. Neurotransmitters from the phrenic nerve may play a role in the migration and development process, much as the testicular descent is influenced by the genitofemoral nerve.

An association between hepatic hypoplasia and neoplasm has been reported.^{2-3 5} These reports do not describe hepatic heterotopia within a diaphragmatic eventration sac continuous with a hypoplastic left lobe of liver as in this case. Hepatic heterotopia in the abdomen or thorax is common, but is extremely rare within a diaphragmatic hernia sac. In this circumstance, heterotopic hepatic tissues has been classified into four main types: (I) a large pedunculated accessory lobe of liver connected by a stalk to the liver; (II) a small sessile accessory lobe of the liver attached to it; (III) ectopic liver with or without connecting to the rest of the liver and (IV) microscopic ectopic liver tissue.¹¹ Types II and III have been reported to occur with diaphragmatic hernia. Our case was type III connecting the heterotopic liver tissue to the mainstream liver.

The operative management of this condition has not previously been described. It was considered unsafe to plicate such a thick, vascular eventration sac. Resection of the eventration could only safely be achieved by including most of the abnormally shaped hypoplastic left lobe of the liver. As the defect was so large with diaphragm absent, repair was achieved by suturing the remaining left lobe of liver and Toldt's fascia with absorbable polydioxanone sutures. The resulting repair was not tense therefore use of an artificial patch was unnecessary.

An intercostal chest drain was inserted as the liver and the eventration sac were vascular structures. Neither bile nor blood drained from the chest, but clearly the baby's ventilation was compromised when the drain was removed prematurely. Increased pulmonary vascular resistance is an almost a universal finding in CDH even when not clinically manifested by right to left shunting at the ductal level. A combination of reactive vasoconstrictive pulmonary hypertension and fixed pulmonary hypertension from lung hypoplasia can make medical management difficult. Management includes lowering of pulmonary vascular resistance and optimising lung recruitment using surfactant, high frequency oscillation ventilation, inhaled nitric oxide or extracorporeal membrane oxygenation.¹²⁻¹⁴ Our case responded very well to the initial treatment requiring a short period of high frequency oscillation ventilation. No studies are available regarding the sequelae and long-term treatment of pulmonary hypertension secondary to CDH.

Learning points

- ▶ Association of hepatic heterotopia and congenital diaphragmatic anomalies is rare.
- ▶ Most cases had deficient pericardium as well whereas our case was exception and phrenic nerve involvement in the embryopathogenesis has been implicated.
- ▶ Association of hepatic hypoplasia and heterotopia with neoplasm dictates long-term follow-up and ultrasound screening.
- ▶ There are four types of hepatic heterotopia described, our case had type III lesion.
- ▶ Association of hepatic and diaphragmatic anomalies makes the repair most difficult and challenging with recurrence rates being higher.

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