
Herniation of a hamartomatous growth of the liver in an infant masquerading as a cardiac tumour

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Introduction

Congenital diaphragmatic hernia is a common birth defect. The incidence varies from 1 in 5000 live births to 1 in 2000, if stillbirths are included. Herniation through the diaphragm could occur through the oesophageal hiatus, the left or right foramen of Bochdalek, the foramen of Morgagni, and communications caused by congenital absence of the left diaphragm or the central tendon [2]. They commonly occur on the left side (75–80%) and occasionally bilaterally [1]. We report an unusual right-sided diaphragmatic hernia, occurring through the central tendon which was initially misdiagnosed as a cardiac tumour.

Case history

A baby girl was born vaginally to non-consanguineous parents at 38 weeks of gestation. Birthweight was 2.3 kg. There was no perinatal complication but she had dysmorphic features comprising microcephaly (OFC 31 cm; <3rd centile; NCHS 2000), polydactyly, cutis aplasia and low set abnormal ears. The cardiovascular system and abdomen were clinically normal. She developed jaundice and shortness of breath on the third

day and was admitted to hospital. Serum bilirubin was below phototherapy level and other investigations excluded septicaemia. 2D+ echocardiography showed hypertrophy of both ventricles with mild systolic dysfunction. Based on echocardiography a diagnosis of mild infiltrative cardiomyopathy was made. She was discharged at the age of 7 days, awaiting the report on karyotyping (karyotyping was normal).

At 10 weeks of age, the child was readmitted with the complaints of peripheral cyanosis and dyspnoea. She was restless. The weight was 3.5 kg; OFC was 34 cm (<3rd centile; NCHS 2000). Early clubbing was present. A hypopigmented skin lesion was noted on the chest. Cardiovascular system was clinically normal. Repeat echocardiography revealed a small ostium secundum ASD. Biventricular hypertrophy and a moderate sized pericardial effusion was seen. A large tumour like mass was seen on the right side of the pericardial cavity, measuring about 3.5 cm × 2.5 cm. CT scan of the thorax was done and a mass lesion was confirmed. While in the ward the child developed generalised tonic seizures. A clinical diagnosis of tuberous sclerosis (TS) was made on the basis of seizures and the hypopigmented skin lesions. The cardiac tumour was presumed to be a

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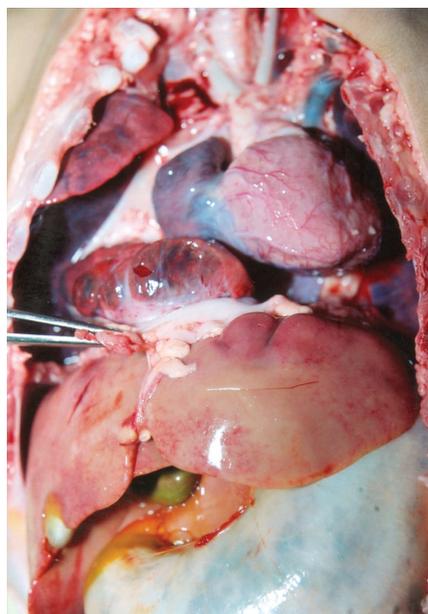


Figure 1. Position of the liver hamartoma in relation to thoracic and abdominal organs, revealed at necropsy examination.

rhabdomyoma. Ultrasound scan (USS) of the brain was done. The echogenicity of the gyri appeared altered with focal ill-defined echogenic areas probably due to tubers. A CT scan of the brain did not detect any abnormalities. USS of the abdomen was done and renal tract abnormalities were excluded. Her cardiac function deteriorated and she died at the age of 12 weeks.

At necropsy a large outgrowth of the right lobe of the liver herniating through the diaphragm and pericardium was seen lying next to the right side of the heart (Figure 1) with a pericardial effusion. The heart was small, probably due to the pressure by the liver mass within the pericardial sac. There was an ostium secundum ASD. Kidneys were macroscopically normal. The outgrowth of the liver showed a few necrotic areas. The histology of the hepatic lesion showed that it was composed of normal liver tissue. Macroscopic examination and histology of brain did not reveal any evidence of TS. No significant pulmonary hypoplasia was noted.

Discussion

Herniation of the liver into the pericardial sac secondary to a defect in the central tendon is the rarest type of diaphragmatic hernia. A literature search revealed fourteen cases [2–9]. All were herniation of a lobe of the liver through the central tendon of the diaphragm into the pericardial sac. Usually they present soon after birth with respiratory distress. However, four cases presented between 1 and 12 month of life [2,9]. With the improvement in ultrasonographic techniques antenatal diagnosis has been possible [3,6].

The present case could be the first in which, although the liver was in its normal anatomical position

a hamartomatous growth originating from it had herniated through the central tendon into the pericardial sac. This would have grown gradually over time as the initial echo did not show an intrapericardial mass.

Initially, based on the clinical features of TS the mass lesion with the pericardium was diagnosed as a rhabdomyoma. Hamartomatous lesions of the liver are known to occur in patients with TS [10]. At necropsy, there were no pathological features suggestive of TS.

This case shows that it is important to include congenital diaphragmatic hernia among the differential diagnosis of a pericardial mass associated with pericardial effusion detected on radiological imaging. It also emphasizes the fact that necropsy examination would help to discover unexpected pathologies, even in the presence of an apparent clinical cause of death.

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