CASE FOR DIAGNOSIS

Congenital lung abnormality in a 1-yr old

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Case history

An asymptomatic 8-week-old male infant was referred for assessment. A prenatal ultrasound had been suggestive of a congenital cystic adenomatoid malformation (CCAM). Following delivery, he had a plain chest radiograph (fig. 1) and went on to have a computed tomography (CT) scan of the thorax (fig. 2), which showed an extrapulmonary mass close to the vertebral column. He proceeded to have magnetic resonance imaging of the thorax to look for intraspinal extension. The appearances were not anticipated, in that the lesion appeared to be intrapulmonary with no extension into the mediastinum, spinal canal or chest wall. The diaphragm was reported as being intact and separating the lesion from the abdominal cavity. There were no abnormal vessels reported. These features were in keeping with an intrapulmonary cystic lesion and the infant was referred for a surgical opinion. As the infant remained well, surgery was deferred until the child was 1-yr old.

Fig. 1. – Plain chest radiograph showing a left-sided abnormality.

Fig. 2. – Thoracic computed tomography scan.

Fig. 3. – Second computed tomography scan.

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was now presumed to be a diaphragmatic hernia was carried out.

At thoracotomy, the mass appeared to be arising from the posteromedial aspect of the diaphragm (fig. 5). The superior aspect seemed consistent with a diaphragmatic hernia, but on further mobilisation the mass appeared to extend through the diaphragm. After opening the diaphragm and exploring the abdomen, it was found to be a mass consisting of diaphragmatic or retroperitoneal tissue extending through a diaphragmatic defect. The 3×3-cm mass was excised and the diaphragmatic defect repaired. Histology of the mass is shown in figure 6.

The boy recovered well and continued to be asymptomatic at 3 months follow-up.

Fig. 4. – Barium swallow.

Fig. 5. – Surgical specimen.

Fig. 6. – Histological specimen.

BEFORE TURNING THE PAGE, INTERPRET THE RADIOLOGY AND SUGGEST A DIAGNOSIS.

Interpretation

Plain chest radiograph

A well-defined mass can be seen through the cardiac silhouette adjacent to the vertebral column.

Thoracic computed tomography scan

There is a ~2-cm-diameter homogeneous (fluid density) mass in the lower left paravertebral region. It appears to be extrapulmonary, but intra-spinal extension cannot be excluded. At the time of this CT the stomach was distended with fluid.

Thoracic computed tomography scan 2

This section shows that at least part of the apparent mass represents herniation of part of the stomach through a diaphragmatic defect.

Barium swallow

There is herniation of the fundus through the left hemidiaphragm.

Surgical specimen

Vascular polycystic structure removed from the diaphragm.

Histological specimen

The entire specimen was composed of lung tissue with surrounding connective tissue capsule. There was no diaphragmatic muscle tissue. Figure 6 shows a central bronchus with cartilage and immature alveoli. There are irregular cystic spaces lined by respiratory bronchiolar ciliated and low cuboidal epithelium within an immature myxoid stroma.

Diagnosis: "Congenital intradiaphragmatic cystic lung abnormality, Stocker's classification type 2"

Discussion

CCAM and bronchopulmonary sequestration are congenital lung masses that are traditionally described as having distinct embryology, pathology and natural history. The term "pulmonary sequestration" was first defined by Pryce in 1946 to describe a disconnected bronchopulmonary mass or cyst with an anomalous systemic arterial supply [1]. SADE *et al.* [2] broadened this description to include a spectrum from abnormal vessels supplying a nonsequestered lung to abnormal pulmonary tissue without anomalous vascular supply. Congenital cystic adenomatoid malformations are hamartomatous lesions characterised by a multicystic mass of pulmonary tissue with proliferation of bronchial structures [3]. This current case appeared to have features of both CCAM and bronchopulmonary sequestration. This association has been reported on several occasions. A study by CONRAN and STOCKER [4] reviewed 46 cases of extralobar sequestration and found that 50% were associated with a coexistent CCAM, all type 2 pattern on histology (Stocker's classification of CCAM

was first published in 1977 and was expanded to five types in 2001). The blood supply to the sequestration in 77% of cases was directly from the aorta [4]. Other studies have reported associations of pulmonary sequestration and CCAM with congenital diaphragmatic hernias and bronchogenic cysts [5–7]. The coexistence of separate anomalies in one lesion suggests that they are ends of a spectrum, rather than separate entities, and indeed it has been suggested that it is more logical to describe all congenital cystic lung lesions as a "congenital thoracic malformation" [8].

Extralobar sequestrations are typically found between the lower lobe and hemidiaphragm; they may also occur in the mediastinum or pericardium and 15% can be found either within or below the diaphragm [9]. In addition to congenital lung abnormalities, the differential diagnoses of diaphragmatic lesions should include neuroblastoma, teratoma, angioma or foregut duplication. These lesions provide a diagnostic challenge. They could be evaluated in the following sequence: chest radiograph, ultrasound with Doppler, followed by CT scan with intravenous contrast or magnetic resonance imaging. These studies should delineate the lesion, its associated vasculature and whether a communication with the tracheobronchial tree exists. An oesophageal contrast study should be considered if there is a history of feeding difficulty [10]. In the case presented here, imaging highlighted a diaphragmatic hernia but did not delineate the mass. Postnatal ultrasound with Doppler was unlikely to have been helpful in delineating the lesion. In some cases, demonstration of the anomalous systemic arterial supply can expeditiously confirm the diagnosis of sequestration [11]. This may obviate the need for preoperative angiography.

The management of prenatally diagnosed sequestration or CCAM in asymptomatic infants is usually delayed elective surgical resection in order to prevent the largely unquantifiable risk of infection, haemorrhage, pneumothorax or malignant transformation. The presence of two or more abnormalities may lower the threshold for surgery.

A rare case of an intradiaphragmatic cystic pulmonary sequestration and the difficulties associated with its diagnosis have been described here. Such "hybrid" cases support the theory that sequestrations and congenital cystic adenomatoid malformations represent a spectrum of anomalies with a common embryological origin and should be described as a congenital thoracic malformation specifying site, arterial supply, venous drainage, histology and connection to foregut or tracheobronchial tree in order to facilitate optimal management.

References

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