



British Paediatric Orphan Lung Diseases (BPOLD)

Name of Disease:

Congenital Lung Malformation

Clinician:

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Clinical details:

Definition

Congenital lung malformations (CLM) are a group of rare congenital malformations that affect the developing lung. These parenchymal lung lesions can be grouped as follows ¹:

- Congenital Cystadenomatoid Malformation (C-CAM)
- Lobar Sequestration
- Bronchial Foregut Malformation aka Bronchogenic Cyst
- Congenital Lobar Emphysema

Congenital Cystadenomatoid Malformation (C-CAM)

The incidence of C-CAM is quoted as 1 in 10,000 to 1 in 35,000 ². This is the most common CLM to be detected on routine antenatal ultrasound screening in the UK ².

C-CAM is characterised by a lack of normal alveoli, as well proliferation and cystic dilatation of terminal respiratory bronchioles. Cartilage is usually absent, and histologically the cystic lesion may be lined by ciliated, cuboidal, or columnar epithelia. Blood supply is normal, and the lesions communicate with bronchial tree. C-CAM lesions vary in size, and can be made up of single or multiple cysts. These lesions are classified according to the system of Stocker ^{3,4}. C-CAM is associated with a risk of malignant transformation ⁵.

Lobar Sequestration

A sequestration is an area of solid, non-functioning lung that has a blood supply originating from the aorta rather than the pulmonary artery, and usually has an absence of communication with the bronchial tree ⁶.

Bronchial Foregut Malformation

Also known as bronchogenic cysts, these can occur early in gestation (producing a central or mediastinal cyst), or later in embryonic development where lesions tend to occur peripherally ⁷. The cysts may be asymptomatic, but can cause respiratory distress as a result of airway compression, a space-occupying effect, or infection.

Congenital Lobar Emphysema

Congenital lobar emphysema (CLE) is a rare congenital lesion of the lung, most commonly diagnosed in the first 6 months of life. This is not usually diagnosed antenally, as the lungs require to be

inflated with air post-natally before the emphysema is manifest. Clinical presentations range from acute respiratory distress in the neonate, to persistent wheezing or chest infections in the older child⁸. Some familial occurrence has been reported⁷, and there is a male preponderance for the condition. In only 25% of cases is bronchial obstruction identifiable as a cause⁷. This ball-valve obstruction causes overdistension of the lung lobe, allowing air entry on inspiration, with collapse of the bronchial lumen in expiration causing air trapping⁹.

Clinical Presentations

The commonest presentation for CLM is by detection of an abnormality on routine antenatal ultrasound scanning. CLM may also be discovered post-natally in an infant presenting with respiratory distress, wheeze, recurrent infections, haemoptysis or pneumothorax.

CLM may also be diagnosed later in childhood, or even in adulthood by presenting as recurrent respiratory infections or even as an incidental chest X-ray finding. The true incidence is unknown, however, as lesions may remain sub-clinical throughout life.

Diagnosis and investigations

An antenatally detected lesion will be monitored by serial antenatal ultrasound scans.

Chest X-ray should be taken in the neonatal period – this often appears normal or shows only minor changes.

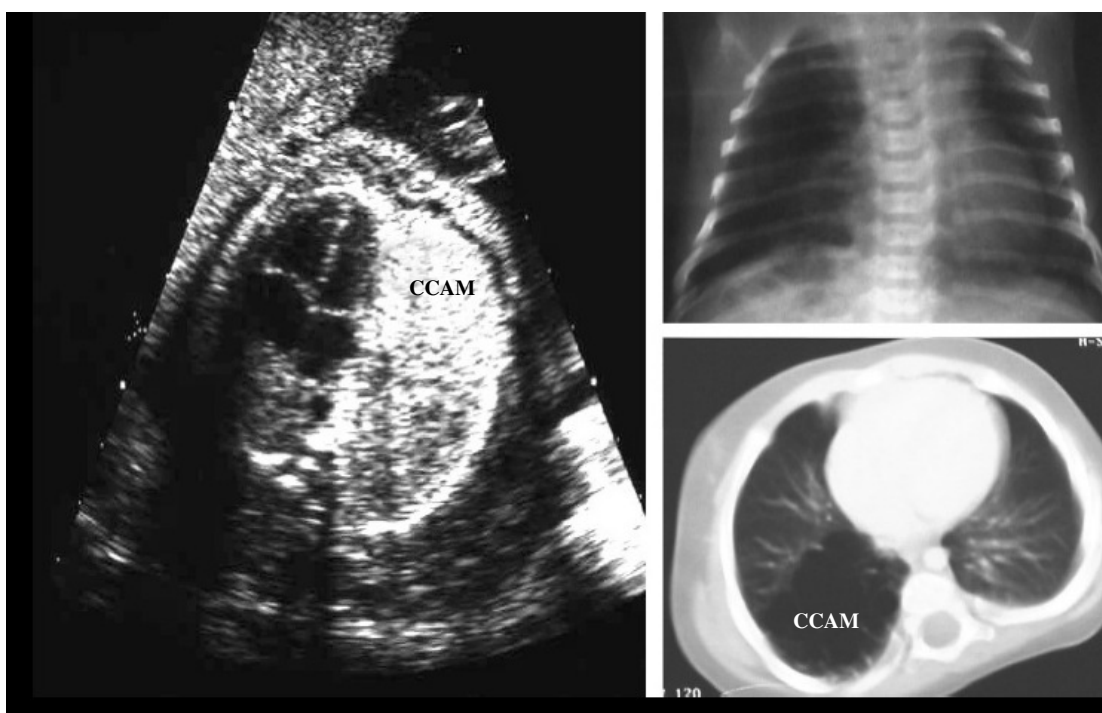
3-dimensional information in the form of a computed tomography (CT) scan of the chest should be undertaken in the first 3 months of life. It is important that CT is undertaken with contrast, in order that blood supply to any lesion can be delineated.

Congenital Cystadenomatoid Malformation (C-CAM)

Even in the event of apparent antenatal regression on ultrasound, it is important that post-natal imaging is carried out (Figure 1).

Figure 1

Presence of an antenatally-diagnosed lesion on ultrasound (A), along with a relatively normal chest radiograph (B), and demonstrable RUL C-CAM on CT scan.



Reproduced from Archives of Disease in Childhood, Lakhoo *et al.* **94**: F73-F76, 2009; with permission from BMJ Publishing Group Ltd.

Lobar Sequestration

This can take the form of an extralobar, or an intrapulmonary sequestration. Around 90% of extralobar sequestrations occur in the left lobar lobe. The lesion is surrounded by its' own pleura and does not communicate with the bronchial tree. Such lesions generally have a systemic (often multiple) arterial supply with venous drainage to either inferior vena cava or the portal system. Such vascular anatomy can cause a significant left to right shunt, although other modes of presentation could be due to infection, compression or merely as an incidental finding on a chest radiograph. Intrapulmonary sequestrations are generally connected to the bronchial tree, have a single feeding vessel, as well as venous drainage to the pulmonary veins⁷.

A CT scan should be performed to assess the extent of the lesion, along with an ultrasound/Doppler or angiographic study (i.e. CT angiography) to delineate vascular anatomy in terms of feeding vessels (arterial supply) and also venous drainage. Such scans should assess aorta and pulmonary arteries and also the diaphragm and liver for transdiaphragmatic feeding vessels. Assessment for other associated congenital anomalies should be undertaken.

Foregut Malformation

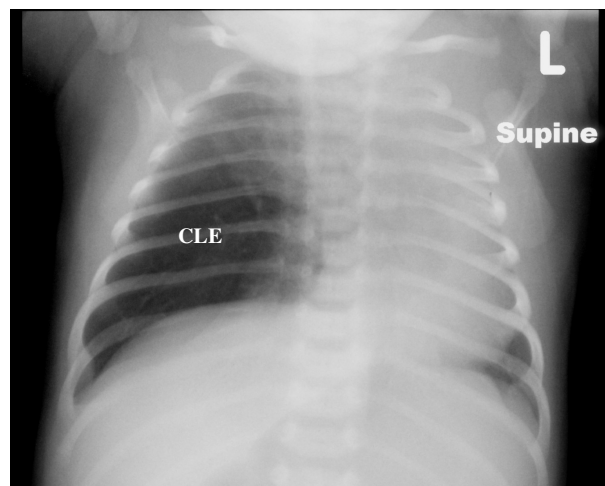
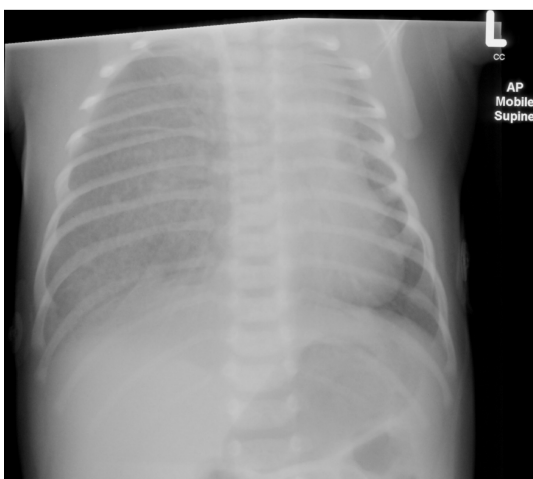
These can be peripheral or central, as described above. Investigations may include CT scan to delineate the size and location of the cyst, as well as whether it appears fluid-filled (i.e. infected). In the cases where airway compression is suggested, a ventilation-perfusion scan may be of benefit in assessing the impact of a cyst upon ventilation.

Congenital Lobar Emphysema

Chest x-ray usually shows a radiolucent lobe and mediastinal shift, with a computed tomography (CT) scan of the chest further delineating the underlying pathology. CLE (Figure 2) is occasionally misdiagnosed as a pneumothorax or isolated bulla, but this can have devastating consequences with attempted aspiration or intercostal catheter insertion. CLE most commonly affects the left upper lobe (40-50%), followed by the right upper lobe (30-40%) and middle lobe (20%)¹⁰.

Figure 2

The initial X-ray (A) was taken aged 5 hours on a baby with tachypnoea and moderate recession. This shows the presence of asymmetric lung fields with mediastinal shift to the left, demarcation of the right middle lobe and fluid in the horizontal fissure. The subsequent X-ray taken at day 13(B), shows lucency of the right middle lobe, with collapse of the right upper lobe and marked mediastinal shift.



A

B

Treatment

Congenital Cystadenomatoid Malformation (C-CAM)

For symptomatic individuals, surgical resection of the C-CAM and surrounding lobe of lung is the general treatment approach, and timing depends on the frequency and severity of intercurrent respiratory infections ².

Controversy exists however, on the optimal management of the asymptomatic post-natally diagnosed C-CAM ^{1,2,5,6,7,12}. Although surgical removal is favoured by many (in view of the potential for malignant transformation within a C-CAM), others would favour a conservative approach to management.

Pro-Surgery:

There is a known potential for malignant transformation within a C-CAM, with pulmonary pleuroblastoma ¹¹, rhabdomyosarcoma ¹³, and bronchoalveolar carcinoma ¹⁴, all having been described in association with C-CAM.

C-CAM can act as a focus for lung infection, resulting in an increased risk of recurrent pneumonia, lung abscess and empyema.

There is a risk of pneumothorax associated with an *in situ* C-CAM ¹⁵, as well as case reports of in-flight air emboli in those with *in situ* C-CAM ¹⁶.

One case series suggests that many patients who present with an asymptomatic C-CAM will subsequently become symptomatic ¹⁷.

There have been cases of intra-thoracic malignancy arising following incomplete C-CAM resection, raising the question of whether surgery should also be considered in those in whom the lesions appear to have regressed either antenatally or post-natally.

Against Surgery:

The incidence of lung malignancy in children is rare.

The excess risk of malignancy attributable to C-CAM is unclear ¹⁸.

There are reports of case series of >100 children in whom conservative management has been associated with no cases of lung malignancy ¹⁹.

Lobectomy is a major thoracic surgical procedure, likely to necessitate a brief intensive care stay, as well as a period of post-operative recovery. The argument for prophylactic surgery would entail this procedure being undertaken before 1 year of age, when anaesthetic risks are greater than in older children. Long-term side effects may be associated with lobectomy.

There have been cases of intra-thoracic malignancy arising following C-CAM resection, either in residual C-CAM tissue, as well as malignancy arising *de novo* in another area of lung ¹¹. This therefore raises the issue that long-term surveillance should be advocated whether lesions have been resected or not; as well as in those in whom a 'disappearing lesion' has been suggested.

A multicentre long-term outcomes study for C-CAM (whether managed conservatively or surgically) is ongoing ²⁰.

Lobar Sequestration

Surgical resection is recommended for the symptomatic patient with a sequestration⁷. It is important that the arterial supply to (and the venous drainage from) the sequestered lung is delineated prior to surgery using angiography or Doppler ultrasound, along with screening for associated congenital anomalies.

Foregut Malformation

Due to the high incidence of infection within bronchogenic cysts, surgical removal is recommended⁷.

Congenital Lobar Emphysema

The symptomatic lobar emphysema requires surgical resection (lobectomy). This condition has no propensity for malignant transformation, and thus in an asymptomatic individual, a period of observation and watchful waiting is appropriate.

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Web links: