Outcome of Congenital Lung Disease

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Introduction

While the various congenital anomalies of the lung are not common, three are of sufficient frequency to come to the attention of most major paediatric thoracic units every year or so. They are congenital lobar emphysema, lobar sequestration and congenital lung cysts. While it has been suggested that these may have a similar embryological origin1), each seems to be a specific clinical entity with its own management problems. Increasing knowledge of their natural history has aided the development of a more rational approach to therapy.

A new congenital lung disease-bronchopulmonary dysplasia is providing concerns to paediatric thoracic physicians. It is a consequence of the treatment of neonatal respiratory disorders with high pressure ventilation and oxygen. The frequency with which it causes chronic lung disease in older children is as yet unclear.

Congenital lobar emphysema

This is an uncommon anomaly occurring in about 1 in 50,000 live births. There continues to be debate about the basic pathology but severe deficiency in bronchial cartilage plates seems to be a characteristic feature2). Usually one or other of the upper lobes are affected but occasionally the disease process involves the middle lobe. It may present first in one lobe and then another. The lobe is uniformly distended but occasionally there are a few cystic areas on its surface.

The hyperinflated lobe frequently causes collapse of the remainder of the lung and displacement of the mediastinal structures. If this is marked in the early days to weeks of life, the infant presents with tachypnoea, breathlessness and often wheezing (Table 1). In this situation there is general agreement that the involved lobe should be removed. This results in resolution of symptoms.

Some times the infant presents beyond the newborn period usually with cough and wheeze. Again, lung compression and mediastinal displacement seems to be the cause of the symptoms.

At times presumed congenital lobar emphysema is an incidental finding on chest x-ray. There is a hyperinflated lobe and some compression of surrounding lung. The problem in these infants and children is to distinguish the congenital lobar emphysema from bronchial atresia.
In the last two presentations there is some debate as to the appropriate therapy. There are those who feel the natural history of patients with less troublesome symptoms is good as often these will resolve with growth\(^3\). They would not recommend surgery. Our approach has been different. Unless there is a specific contraindication, we would normally recommend surgery in all infants and children with congenital lobar emphysema. Three factors influence this approach. The first is we have seen tension develop many years after initial presentation with minimal symptoms. This has resulted in marked mediastinal displacement and the development of a chest wall deformity. A second problem is the inability to distinguish absolutely lung cysts, bronchial atresia and congenital lobar emphysema. There is good evidence that all congenital lung cysts should be removed (see below) and the same also seems true of bronchial atresia as infection is likely to develop beyond the atretic bronchus\(^4\). We have also seen infection develop in an emphysematous lobe. The final reason is that children with a congenital lobar emphysema have to remain under medical supervision and probably require annual chest radiographs. In the hands of an experienced paediatric thoracic surgeon and paediatric anaesthetist, the morbidity from lobectomy for this disorder should be very small. Therefore our view is that the risk of surgery is less than the risk of doing nothing in these infants and children with minor or no symptoms.

There is some evidence that children who have had a congenitally emphysematous lobe removed in infancy will have airways disease in later childhood. Two studies have shown some minor reduction in forced expiratory flows in a proportion of such children\(^5,6\). Usually there are no symptoms but occasionally there may be some minor wheeze. The reasons for this are unclear but it seems probable that the cartilage deficit is present in other parts of the lung but probably in a less severe form.

**Congenital cystic lung disease**

There probably are a group of developmental anomalies that can result in intra-pulmonary cysts. A variety of nomenclatures have been suggested for these including cystadenomatoid malformation, alveolar cysts and bronchogenic cysts\(^6\). From a natural history and management point of view separating them into various entities does not seem particularly helpful. They are probably about twice as frequent as congenital lobar emphysema and so occur in about 1 to 25,000 births.

Intra-pulmonary cysts will almost certainly eventually produce symptoms. If they are large and particularly if their bronchial communication is subject to a ball valve effect, they can produce an area of tension within the lung. This results in compression of normal lung and mediastinal displacement. The symptoms are those of respiratory distress-tachypnoea, breathlessness and wheezing (Table 2). This is a common mode of presentation in the early days or weeks of life but occasionally tension will develop in a pre-existing cyst quite suddenly in later childhood or adolescence.
Because the cyst has abnormal communication with the bronchus, it is susceptible to infection. Thus, presentation with infection is common. Of course, there will often be a debate as to whether the cyst has arisen as a result of a destructive pneumonia such as staphylococcal infection or as infection in a congenital cyst. The clinical and radiological pattern may be suggestive—a multi-loculated cyst in general is more likely to be congenital in origin. True acquired cysts will resolve within 6–9 months whereas infection is likely to recur in a congenital cyst. If the possibility is that the cyst is acquired, then a period of observation for 6–9 months would be justified.

Pneumothorax is an uncommon disorder in children and early adolescence. However, our experience has been that provided there is not some underlying lung disease such as asthma or cystic fibrosis, pneumothorax in the paediatric age group is almost always due to rupture of a sub-pleural cyst. Whether these are truly congenital in origin or arise during the period of rapid growth in early adolescence is unclear. Nevertheless the pneumothorax is likely to be recurrent if the local area of lung dysplasia is not over sown or removed.

A few lung cysts have been found incidentally on chest radiographs. However, two of our patients who presented in this way were not subjected to surgery at the time of presentation, subsequently developed infection in the cyst.

It is our practice to recommend surgical removal of all congenital intrapulmonary cysts at the time of diagnosis because they are very likely to develop complications. Usually the cyst can be removed by segmental resection but occasionally a lobectomy is required. If cysts involve more than one lobe, then segmental resection should be the approach.

There are no long term followups of patients who have had lung cysts removed. Our impression is that at least some seem to have ongoing airways disease. Perhaps there is some relationship between intrapulmonary cysts and congenital lobar emphysema. Certainly the bronchi supplying some congenital intrapulmonary cysts has defective cartilage plates.

**Intralobar sequestration**

While it has been questioned as to whether intralobar sequestration is a specific entity, the evidence strongly supports that it is. This anomaly is best defined as an area of lung which is a result of congenital maldevelopment, lacks normal bronchial communication, has its blood supply directly from the aorta or one of its branches with venous drainage either into the systemic or pulmonary system. It typically is found in the postero-medial part of the left or right lower lobe. The systemic arterial supply classically comes through the diaphragm.

Its frequency is similar to that of congenital lobar emphysema—about 1 in 50,000 live births. Because the area of lung lacks normal bronchial communication, it is at risk of developing infection or tension from a ball valve effect.
Presentation with infection is the common one (Table 3). This occurs more frequently in later childhood and adolescence. This somewhat late presentation has been one reason why the congenital nature of the lesion has been questioned. Typically the area of lung infection is multicystic and its site in the postero-medial part of a lower lobe should strongly suggest the diagnosis. If the cause of the infection is not recognised it will be slow to resolve or recur. Surgical removal, preferably by segmental resection, is the appropriate treatment. This may need to be deferred for some days if an the acute infection requires treatment with antibiotics. However, the infection will not resolve completely without removal of the abnormal lung.

Breathlessness from birth is another common presenting feature. Two features of the intralobar sequestration can cause breathlessness. First, tension can develop within the abnormal lung leading to displacement of normal lung and of the mediastinum. In this way the effects of sequestration are very similar to those of a congenital lung cyst or even congenital lobar emphysema. Because the sequestration has an aortic blood supply, it can act as an arteriovenous shunt. If the blood flow through it is high, this can compromise the circulation and result in heart failure. Treatment of the former group is surgical removal but in the latter group embolisation of the feeding artery may be sufficient to control the symptoms. We have been following this latter procedure for more than 5 years and subsequent problems have not developed in the area of lung supplied by the embolised vessel.

Patients presenting with the incidental radiological finding of an intralobar sequestration should have this removed as infection or tension will develop.

There is still some argument about the appropriate pre-operative investigation. Usually an aortogram is necessary to confirm the diagnosis and this provides important information for the surgeon in planning his exploration. If the surgeon is not aware of the possibility of a large feeder artery transversing the diaphragm, a surgical disaster may result from injudicious division of the artery.

There have been no long term followup studies on children who have had an intralobar sequestration removed. However, our impression is that these patients remain very well and have no further problems.

**Outcome of congenital lung remission**

Thus the natural history of congenital lobar emphysema, lung cysts and lobar sequestration is eventually to result in clinical symptoms. They are not benign lesions and the outcome is likely to be unfavourable unless they are removed. What is still unclear is whether particularly congenital lobar emphysema and lung cysts are localised anomalies. There at least is some evidence to suggest that patients with them can have residual airways disease. Long term followup of patients successfully treated by surgery will be necessary to determine the frequency of this.
Bronchopulmonary dysplasia

No discussion of the outcome of congenital lung disease would be complete without making some reference to the new congenital lung disease-bronchopulmonary dysplasia. This lung damage resulting from the treatment of neonatal lung disease, particularly but not exclusively, hyaline membrane disease in premature infants.

The early natural history of bronchopulmonary dysplasia seems to be either death within 6-9 months or alternatively progressive improvement by 12-18 months. Very few infants remain oxygen dependent after the age of 18 months.

There are now long term followup studies which are giving conflicting results. A study from Boston Children’s Hospital suggested that airways disease was frequent in children aged 8-11 who had had bronchopulmonary dysplasia\(^9\). They had symptoms of cough, wheeze and breathlessness and if anything the disease had progressed between 8 and 11.

Our own followup of a group of very small premature infants from one neonatal intensive care nursery is more reassuring. Only a small number had physiological evidence of airways obstruction. Further long term followup will be necessary to determine the frequency of chronic lung disease in survivors of neonatal respiratory disorders treated by mechanical ventilation and oxygen.

References

Table 1
Mode of Presentation of 32 Patients with Congenital Lobar Emphysema

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<td>Wheeze</td>
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<tr>
<td>Cough</td>
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<td>Incidental radiological finding</td>
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Table 2
Mode of Presentation of 62 Patients with Congenital Intra-Pulmonary Cysts

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<tr>
<td>Infection</td>
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<tr>
<td>Pneumothorax</td>
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<td>Symptomless</td>
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<td>Chest wall deformity</td>
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Table 3
Mode of Presentation of 27 Patients with Intraloberal Sequestration

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