with relatively minor degrees of overtreatment and the new assay methodology will allow further evaluation of this aspect of congenital hypothyroidism.

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Pitfalls in the diagnosis of congenital diaphragmatic hernia

Congenital diaphragmatic hernia (CDH) is a rare anomaly occurring in one in 3600 live births. If affected fetuses are taken into account the incidence is increased to one in 2200.1 An increasing proportion of these cases are being diagnosed as a result of routine antenatal scanning. There remain a significant number of affected patients who present for the first time in the neonatal period and a further group presenting as older infants or children.

Neonatal diagnosis and pitfalls

The classical presentation of a neonate in respiratory distress, with a scaphoid abdomen, apparent dextrocardia, and multiple air and fluid filled bowel loops in the left hemithorax on chest radiography presents no diagnostic difficulty. Occasionally the chest radiograph is taken before the passage of swallowed air into the herniated intestine and will thus demonstrate a radio-opaque hemithorax. The absence of intestinal gas in the abdomen should provide the diagnostic clue in these cases.2 In all cases the abdomen should be included on the radiograph if CDH is suspected. The existence of a previously normal chest radiograph does not preclude the diagnosis.3 The defect in the diaphragm may be plugged by a solid abdominal viscous, for example, liver, spleen, or omentum.4 The delayed appearance may on some occasions be related to initially raised intrathoracic pressure which when lowered allows the abdominal viscera to enter into the affected hemithorax.5

There is a further group of neonates with delayed appearance of right diaphragmatic hernia associated with group B streptococcal infection, and neonates with relapsing streptococcal infection should have this diagnosis positively excluded.4,6 Right sided hernias may present as a pleural effusion, the obstructed hepatic venous outflow from herniated liver causing vascular congestion and transudation from the liver surface.4,8 The effusion may compress the adjacent lung resulting in atelectasis which may mimic primary pulmonary pathology, or may prevent identification of either the herniated liver or the diaphragm itself. Ultrasound should establish the diagnosis in these cases. Paraoesophageal and oesophageal hiatal hernias are generally associated with vomiting rather than respiratory embarrassment. Large paraoesophageal hernias are usually identified in the posterior right cardiophrenic region and easily confirmed on barium studies. The differentiation of cystic adenomatoid malformation, congenital lobar emphysema, and CDH may present diagnostic difficulty as they may all present with respiratory distress at or shortly after birth.9-11 If the chest radiograph is taken too early the affected lung in cystic adenomatoid malformation and congenital lobar emphysema may be fluid filled. There are several diagnostic pointers which assist in establishing the correct diagnosis. The presence of normal intestinal gas shadows within the abdominal cavity should raise doubts about the diagnosis of CDH. The position of the tip of the nasogastric tube may be helpful when the stomach lies above the diaphragm, and in these instances it may be a prognostic indicator, as the presence of an intrathoracic stomach has been equated with an adverse prognosis.12 On rare occasions air or contrast may be instilled via the nasogastric tube to determine the position of the stomach and small intestine. Ultrasound may prove helpful and may demonstrate the diaphragmatic defect with bowel or, if on the right, liver passing through. Congenital lobar emphysema usually involves the upper lobes and the chest radiograph most commonly shows hyperlucency rather than an appearance similar to bowel gas shadows.9 There have been reports of the other congenital bronchopulmonary malformations being misdiagnosed as CDH, but these reports stress the importance of careful interpretation of the chest and abdominal radiographs, and if necessary, the use of ultrasound.13,14

Eventration is more common on the right, usually contains liver, and is most easily diagnosed by ultrasound. Occasionally the fine distinction between a partition containing a few strands of muscle, that is, a diaphragmatic eventration, and a peritoneal sac without striated muscle, that is, a hernia, may prove difficult even at surgery, but in such cases the distinction is unimportant.15

Staphylococcal pneumonia that has progressed to produce pneumatoceles may have an appearance on chest radiography that on first inspection mimics CDH.16 Staphylococcal pneumonia may affect either lung and the bowel gas pattern will be normal. The clinical presentation will be different and the presence of earlier films should prove helpful.

Urgent surgery for CDH has been superceded by a period of preoperative resuscitation and stabilisation which may extend over several days.17 The rapid presumptive diagnosis of CDH remains essential for the correct management to be instituted. The period of preoperative stabilisation allows ample time for the definitive diagnosis to be established by the full range of necessary radiological investigations if required.

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Diagnosis and pitfalls in the older child
The initial impression that CDH is rare beyond the neonatal period has been revised. The reported incidence ranges from 5 to 25% of patients with CDH. The wide variety of symptoms and signs in the older age group compound the diagnostic difficulty. Recurrent chest infections, acute respiratory distress, recurrent vomiting, diarrhea or constipation, and failure to thrive are some of the reported presenting symptoms. The clinical signs may be equally non-specific. The hernias may be intermittent or only demonstrable in Trendelenberg’s position. Thus the presence of a normal or previously normal chest radiograph does not exclude the diagnosis. At least 22 documented cases of left posterolateral hernia have been reported in the English literature in patients with previously normal chest radiographs. The size of the hernia bears no relation to the severity of the symptoms. The most frequent misdiagnosis after chest radiography appears to be pneumonia, although other diagnoses such as pneumothorax, cystic lung bullae, and gastric volvulus have been reported.

Careful interpretation of the chest and abdominal radiographs at presentation will, as in the neonatal group, usually lead to the correct diagnosis. The presence of haustral markings or valvulae conniventes will help identify bowel, although these may be difficult to appreciate on a supine film and an erect film may be required. The passage of a nasogastric tube may enable the correct diagnosis to be made in left sided hernia when the stomach lies in the chest. Other imaging modalities which may be helpful include a barium meal and follow through or barium enema as most hernias in this group will contain some part of the gastrointestinal tract. Recent reports suggest that Morgagni hernia more frequently contain bowel than was previously recognised. Although Morgagni hernia are more likely to present as a chance finding, the presence of a hollow viscus increases the likelihood of symptoms. As in neonates, right sided hernias frequently contain colon and in the older child may be asymptomatic. Ultrasound may demonstrate the diaphragmatic defect, if required. In both left and right sided herniations intestinal obstruction or strangulation is a well reported occurrence.

In both groups the correct diagnosis is nearly always apparent on close inspection of the presentation radiographs. The most likely diagnostic error is cystic adenomatoid malformation in the neonatal group and pneumonia in the older group. If further investigation is required the passage of a nasogastric tube may prove helpful in both groups. If the diagnosis is still not clear an ultrasound in the neonatal group and a barium study in the late presentation group will establish the diagnosis in most instances.

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