CLINICAL QUIZ (p310) ANSWER

What is the diagnosis?

The middle and lower zones of the left chest are filled with bubbly shadows without fluid levels. The left hemi-diaphragm is not well visualised. The gastric bubble is absent. Cardiac apex is obscured. There is also mild scoliosis. The radiological features are compatible with left sided diaphragmatic hernia. The diagnosis was confirmed by CT thorax and the defect was successfully repaired by thoracoscopic technique.

What is late presenting diaphragmatic hernia?

Diaphragmatic hernia is a condition where a defect in the diaphragm results in herniation of abdominal viscera into the thoracic cavity. The majority of diaphragmatic hernias are congenital in nature, known as congenital diaphragmatic hernia (CDH). In contrast, acquired diaphragmatic hernias are due to trauma and are less common. The underlying pathophysiology of CDH is believed to be due to the persistence of the posterolateral pleuroperitoneal canal in the diaphragm which allows abdominal viscera to herniate into the thoracic cavity.[1,2]

CDH can be diagnosed prenatally with ultrasonography. Cases without prenatal diagnosis commonly present shortly after birth with acute respiratory distress and the classical "scaphoid abdomen", as a result of pulmonary hypoplasia and pulmonary hypertension associated with abdominal visceral herniation. Less than 3% of the patients remain undiagnosed within the neonatal period.[3] This is believed to be due to a milder degree of visceral herniation, lesser compression onto the lungs and absence of pulmonary hypoplasia.[4] Another possible reason for late presentation is thought to be due to bowel herniating through a diaphragmatic defect that has previously been occluded by the spleen and liver during early period of life.[4] These patients may present anytime beyond the neonatal period and are called late-presenting CDH.

Clinical presentations of late presenting CDH

Compared to early presenting CDH where the diagnosis is usually straightforward, the symptoms are usually more vague and less acute in late presenting CDH.[5] A study on late presenting CDH suggested three types of presentation: acute respiratory and gastrointestinal symptoms, chronic nonspecific symptoms such as abdominal pain and constipation, and lastly, incidental discoveries in asymptomatic patients.[5]

Clinicians most often use chest radiographs to help guide diagnosis. To confirm the diagnosis, additional imaging including abdominal radiographs, upper and lower gastrointestinal contrast studies and CT thorax with contrast may be necessary.[6] However, owing to the variable clinical presentations as well as the low incidence of congenital anomaly in older children, there is often a delay in diagnosis and frequent misdiagnosis. The commonest initial misdiagnosis is pneumonia.[6] Furthermore, studies have found that 25% of initial chest radiographs were misinterpreted as tension pneumothorax or pyopneumothorax.[7]

Complications of CDH

Complications of late CDH occur in around 10% of patients most commonly with both large bowel strangulation and necrosis as well as gastric volvulus. There are also reported cases of gastric perforation due to strangulation.[8] Misdiagnosed cases treated as pneumothorax or pleural effusion may also suffer from iatrogenic complications during chest drain insertion leading to gastric perforation.[3]

Treatment

All patients with CDH require surgical intervention to repair the defect on the diaphragm. Prognosis of a late presenting CDH is generally excellent but delayed diagnosis may significantly increase morbidity.[3] Clinicians should always include congenital lesions in their differential diagnoses if the clinical presentations are recurrent or atypical.

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