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*CASE FOR DIAGNOSIS*

# An infant with respiratory distress and failure to thrive

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## Case history

A 4-month-old male was referred to the Dept of Paediatric Surgery at the authors' institution, for the investigation of respiratory distress, failure to thrive and complete situs viscerum inversus since birth. A chest radiograph obtained a few hours after birth showed situs viscerum inversus, left apical atelectasis, hyperinflation and hyperlucency of the remaining left lung, and a mild shift of the mediastinum toward the right. The right lung was normal. The infant was born at term, after an uneventful pregnancy, by elective caesarean section. The parents were consanguineous and the baby had a positive maternal family history for situs viscerum inversus and chronic respiratory diseases.

On admission, the infant's general condition was poor, his body weight was 5.490 kg (5th percentile) and he was 62.5 cm in height (10th percentile). Clinical examination disclosed tachypnoea (70 breaths·min$^{-1}$), chest retractions and barrel chest. The arterial oxygen saturation in room air was 90% when the child was awake, reaching 97% with 2 L·min$^{-1}$ of oxygen. The cardiac frequency was 150 beats·min$^{-1}$. On examination of the chest, auscultation revealed diffuse bilateral expiratory wheezing and right posterobasal crackles. The remainder of the examination was compatible with situs viscerum inversus and negative. Cardiac evaluation (electrocardiogram and echocardiogram) was compatible with dextrocardia. Arterial blood gas analysis at room air showed an arterial oxygen tension of 6.8 kPa (51 mmHg) and an arterial carbon dioxide tension of 5.7 kPa (43 mmHg). Red blood cells were found at a level of $5.05\times10^{12}$·L$^{-1}$, haemoglobin at 14 g·dL$^{-1}$, white blood cells at $12.6\times10^{9}$ L$^{-1}$ (neutrophils 37%, eosinophils 3%, basophils 2%, lymphocytes 51%, and monocytes 7%), and the erythrocyte sedimentation rate at the first hour was 13. C-reactive protein, immune profile, blood chemistry, and sweat test were normal. Microbiological studies for common bacteria, *Mycoplasma pneumoniae*, *Chlamidia trachomatis*, respiratory viruses, cytomegalovirus, and Epstein Barr virus, were negative. A chest radiograph was repeated (fig. 1). Flexible bronchoscopy showed a slightly right-shifted trachea and a normal inverted bronchial tree, without signs of inflammation or mucus plugs. The child also underwent perfusion (fig. 2) and ventilation lung scintigraphy, with measurement of the ventilation/perfusion ratio ($V'/Q'$ ratio), and virtual computed tomography (CT) scan (fig. 3). The $V'/Q'$ ratio calculated from the lung scan showed a moderate mismatch involving the right upper lung ($V'/Q'$ ratio: 2.2–2.3), with a normal right lower lobe ($V'/Q'$ ratio: 0.86–1.08) and left lung ($V'/Q'$ ratio: 0.85–1.04).

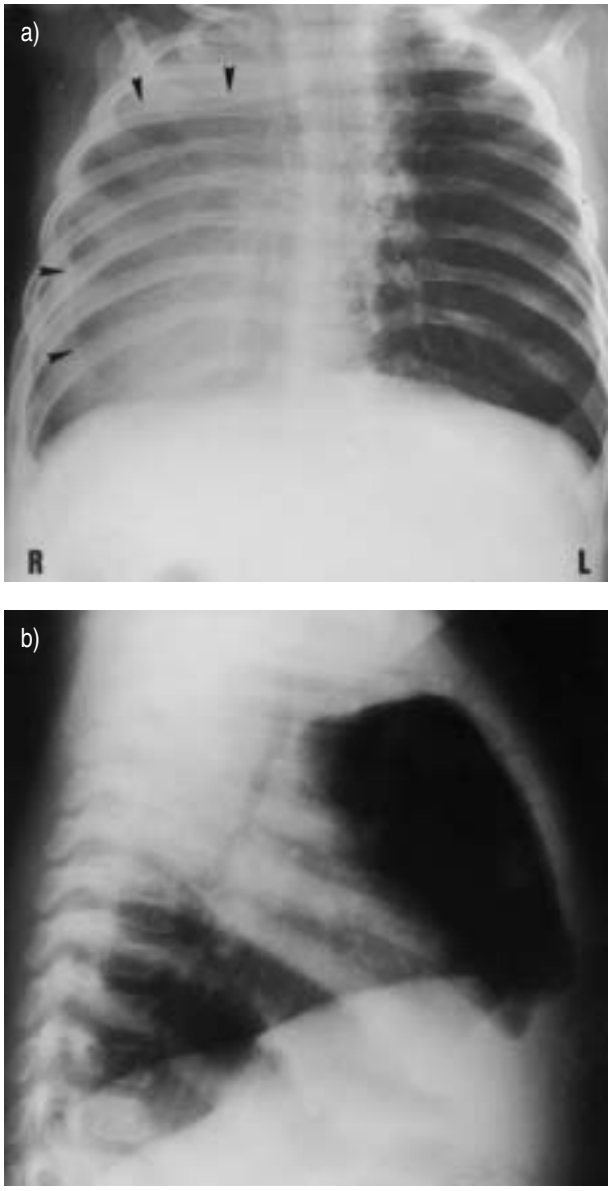


Fig. 1.–a) Anteroposterior and b) lateral chest radiograph.

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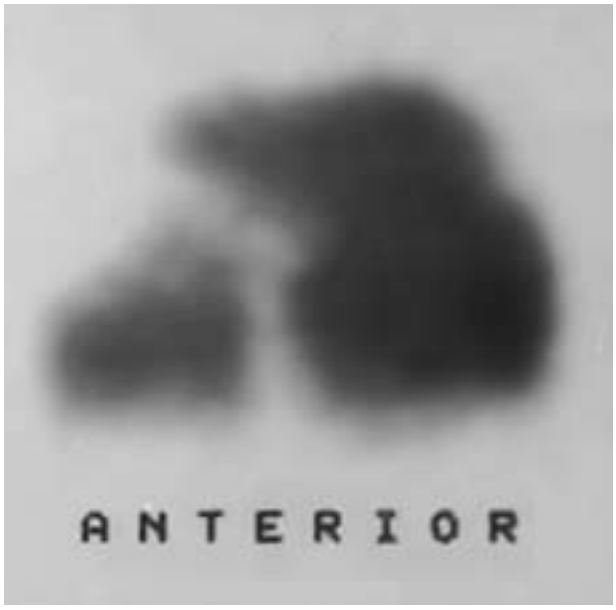


Fig. 2. – Lung perfusion scintigraphy.

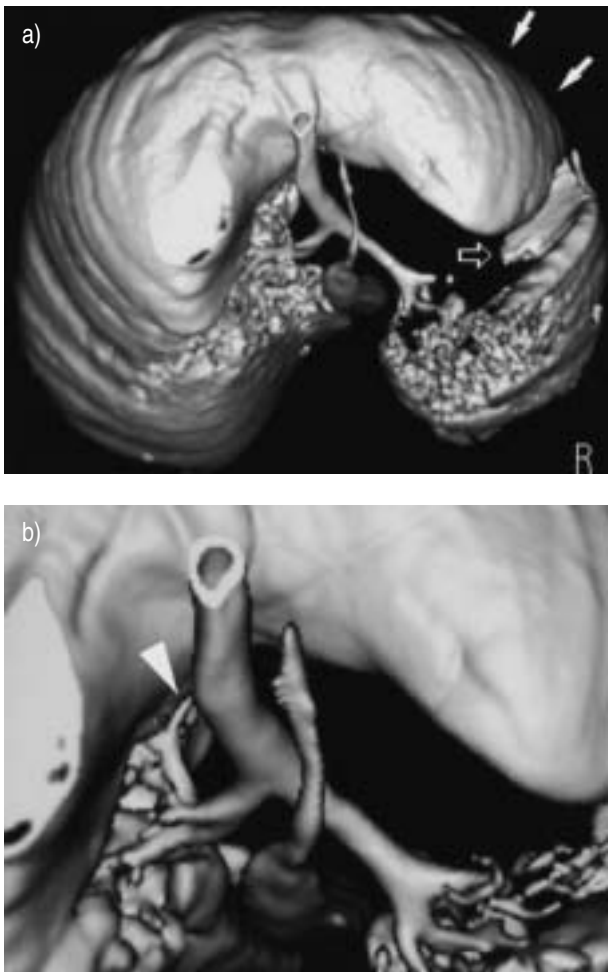


Fig. 3. – a) Thorax computed tomography scan with a three-dimensional model. b) Magnification. The lungs are seen from behind.

**BEFORE TURNING THE PAGE, INTERPRET THE CHEST RADIOGRAPH (FIG. 1), LUNG SCINTIGRAPHY (FIG. 2) AND CT SCAN (FIG. 3) AND SUGGEST A DIAGNOSIS.**

### Interpretation of the chest radiographs, lung perfusion and computed tomography scans

Herniation of the left lung across the anterior superior mediastinum (arrows), reaching the anterolateral right thoracic wall is observed (fig. 1). There is posterior displacement of the heart and the left apical atelectasis is no longer detectable. There is also right apical lung opacity, probably due to right upper lobe compressive atelectasis.

Hypoperfusion of the superior lobe of the right lung is shown in figure 2. There is also normal perfusion of the left lung, which is hyperexpanded and herniates across the superior mediastinum.

Figure 3 shows a hyperexpanded left lung parenchyma that herniates across the anterior superior mediastinum (solid arrows), reaching the anterolateral right thoracic wall. The right upper lobe appears to be compressed and pulled downward (open arrow). The bronchus for the medial segment of the middle left lobe is stretched to the right, behind the trachea (arrow head), and reaches the right hemithorax.

### Diagnosis: "Complete situs viscerum inversus, primary ciliary dyskinesia, and congenital lobar emphysema of the left middle lobe with herniation of the left lung into the right hemithorax"

### Treatment and clinical course

This infant had situs viscerum inversus, congenital ultrastructural anomalies of the cilia, chronic respiratory distress and failure to thrive. It was suspected that the child had congenital lobar emphysema (CLE) of the left middle lobe. This defect had shifted the mediastinum to the right, thus compressing the ipsilateral and contralateral upper lobes, and causing chronic respiratory distress.

The child′s initial clinical conditions were stable and not unduly severe. Imaging studies were not conclusive either for lung hypoplasia (suggested by perfusion lung scan) or CLE (supported by chest radiography and CT scans). Because the mismatched defect in the upper and middle right lobes, as shown by measurement of the $V'/Q'$ ratio, seemed an unlikely cause of the patient′s hypoxaemia, a "wait and see" strategy was decided on, with chest physiotherapy and medical treatment. After 3 months of follow-up, respiratory distress and chronic hypoxaemia worsened, and the infant was sent to surgery to remove the emphysematous lobe, which, in the authors′ opinion, was the probable cause of the child′s respiratory distress.

At operation it was found that the left middle lobe was hugely hyperinflated, projecting to the right hemithorax, and collapsing the left and right upper lobes. Standard left middle lobectomy was performed with immediate partial expansion of the left and right upper lobes. At gross examination, the emphysematous left middle lobe had peripheral areas of hyperinflation, and microscopy showed focal haemorrhage, alveolar distention and rupture, consistent with CLE.

The postoperative course was uneventful and the child was discharged on day 12. At programmed re-admission, 20 days after surgery, the child′s clinical condition had improved remarkably, with complete remission of respiratory distress. Peripheral $O_2$ saturation was >95% at room air, even during sleep. A chest radiograph, obtained 1 month after surgery, showed re-expansion of the previously collapsed left upper lobe, with persistence of the transmediastinic hernia and consolidation of the right upper lobe. The repeat perfusion lung scan showed that the function of both the right lung and left upper lobes had improved.

## Discussion

Primary ciliary dyskinesia (PCD) is a term designating a group of congenital diseases, involving impaired motion of the cilia [1]. Its prevalence has been estimated to be between 1:20,000 and 1:30,000 [2]. Clinical findings in patients with PCD include a combination of the following: situs viscerum inversus, chronic rhinitis, nasal polyps, sinusitis, chronic cough, bronchitis, bronchiectasis, and male infertility. The respiratory tract manifestations are related to a reduced mucociliary clearance secondary to the abnormal motility of respiratory cilia.

In this case, the patient had situs viscerum inversus and respiratory distress developed soon after birth, in an apparently normal baby. Owing to the symptoms and the family history for situs viscerum inversus and chronic respiratory diseases, PCD was suspected. This diagnosis was confirmed by ultrastructural studies of the respiratory cilia. Diagnosis of PCD is difficult in newborns. Nonetheless, PCD should always be suspected in term neonates with respiratory distress or pneumonia and no obvious predisposing risk factors, in neonates with chronic purulent rhinitis, and in those with defects of laterality or complex heart disease [2, 3]. It should also be considered in neonates with respiratory problems and diagnosis of oesophageal atresia, biliary atresia, hydrocephalus, polysplenia or a family history of PCD [2].

In the present patient, the clinical and imaging findings suggested three possible diagnoses: right and left upper lobe atelectasis with secondary emphysema of the left lung; right upper lobe hypoplasia and compensatory left lung emphysema; or primary left lung emphysema and compressive atelectasis of the right and left upper lobes.

Newborns and infants with PCD and respiratory distress may present with radiological findings of atelectasis, secondary to mucus plugs, with or without compensatory hyperinflation of the remaining pulmonary parenchyma [4]. In this patient, the clinical, endoscopic, and radiological findings excluded this possibility. In the authors′ opinion, atelectasis of the upper lobes alone was not sufficient to justify the severity of respiratory distress. In addition, flexible bronchoscopy demonstrated a normal bronchial tree without any inflammatory signs or mucus plugs. Yet, the possibility of mucus plugs during pregnancy or the newborn period could not be excluded.

Although the perfusion scan suggested the presence

of right upper lobe hypoplasia, the radiological, bronchoscopic, and CT scan findings did not. The left apical atelectasis seen on the very first chest radiographs, disappeared on films taken a few months later. In addition, bronchoscopy and CT scan clearly demonstrated that the bronchial tree was reversed but otherwise normal. CT scan disclosed the right upper lobe compressed by the hyperinflated left middle lobe, herniating to the right.

The only possible diagnosis was a CLE of the left middle lobe with compressive atelectasis of the right and left upper lobes, as suggested by CT scan and by the serial chest radiographical findings. CLE is a life-threatening respiratory disorder presenting in infancy. It is characterised by overdistension of one or more of the lobes of the lung caused by trapping of air within the affected lobe, followed by the inability of the affected lung to deflate normally. Congenital deficiency of bronchial cartilage is the most common cause of CLE. Less common causes include intraluminal bronchial obstruction, bronchostenosis, bronchial torsion, bronchial mucosa thickening, obstructing mucosal flaps, cartilagineous septae, bronchial atresia, and extrinsic bronchial compression [5].

In the present patient, the only imaging test incompatible with lobar emphysema was the perfusion lung scintigraphy. The $V'/Q'$ lung scan, however, appears to elucidate the functional rather than the anatomical status of lung parenchyma, showing only aspecific features not exclusive to CLE [6]. $V'/Q'$ lung scintigraphy is therefore more helpful in deciding whether or not to operate on the involved parenchyma than in differentiating between primary and secondary emphysema.

The differential diagnosis between atelectasis with compensatory contralateral hyperinflation and CLE compressing the adjacent lobes is crucial for the therapeutic approach. In patients with PCD and atelectasis, surgery allows only transitory improvement of clinical conditions and the appropriate treatment is regular chest physiotherapy and antibiotics [2]. By contrast, in patients with CLE and severe respiratory distress, prompt surgery is indicated [7]. The natural history of CLE is often progressive and includes potentially life-threatening respiratory insufficiency from compression of the adjacent normal lung. Lobectomy is the procedure of choice, with excellent long-term clinical results [8].

To conclude, pulmonary emphysema has been described in older children and adults with primary ciliary dyskinesia, but to the best of the authors' knowledge, this is the first case of primary ciliary dyskinesia and associated congenital lobar emphysema in a newborn. In newborns with primary ciliary dyskinesia, respiratory distress, and radiological signs of lung hyperinflation, congenital lobar emphysema should be ruled out.

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