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Clinical Problems

Asymptomatic Pulmonary Agenesis: Our Experience with Two Cases

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Abstract

Pulmonary agenesis, aplasia and hypoplasia are very rare major congenital anomalies. In the great majority of these conditions, diagnosis is usually made at or soon after birth. However, extremely rare asymptomatic cases may go unnoticed until adulthood. We present two asymptomatic patients with pulmonary agenesis in whom the diagnosis was delayed until their fourth and fifth decades, respectively. In both patients, ultimate diagnosis was coincidentally made based on abnormal appearance on chest X-rays obtained for other symptoms. Contrast-enhanced thorax CT and MRI angiography were found to be the most useful tools in their management.

Keywords: pulmonary agenesis, MRI angiography

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INTRODUCTION

Congenital pulmonary anomalies are seldom reported. The widely accepted description of major anomalies was made by Boyden in 1955, as agenesis, aplasia and hypoplasia [1]. Due to early onset of the symptoms for obvious reasons, diagnosis of agenesis and aplasia is usually made soon after birth [2-5]. Conversely, some hypoplasia cases may go unnoticed and the diagnosis may be delayed until adulthood [6,7]. Pulmonary agenesis diagnosed in adulthood is an extremely rare condition, and only a few cases have been reported in the literature [8-10].

In this article, we report two cases with pulmonary agenesis that had remained totally silent until the fourth and fifth decades.

CASE REPORTS

Case 1

A 43-year-old male patient was admitted to our hospital with the complaints of dyspnea, cough, sputum production and edema in lower extremities. The patient stated that he had no prior pulmonary complaint or physical restriction until 10 days before the admission. Physical examination revealed no breathing sound on the left hemithorax and inspiratory rales on the right basal hemithorax. His posteroanterior (PA) chest X-ray (Figure 1) demonstrated an elevated left hemidiaphragm, complete opacification of the smaller left hemithorax, left-sided shifting of mediastinal and tracheal structures, and right basal heterogeneous infiltration. In laboratory analysis, leukocytosis (15000/mm^3), hypoxemia (PaO_2= 45 mm Hg) and restrictive type respiratory pattern were detected (FVC=63% predicted, FEV_1=53% predicted, FEV_1/FVC=72%). In bronchoscopic examination, left main bronchus was found to end just 2 cm after the main carina. In addition, the patient had multiple genetic anomalies: short left arm, small left hand, small ipsilateral facial characteristics and right pelvic kidney. His contrast-enhanced thorax computed tomography (CT) revealed total absence of left pulmonary parenchyma and left pulmonary artery (Figure 2). The patient was thus diagnosed as having left-sided pulmonary agenesis. His acute condition was attributed to pneumonia on the right side with accompanying right heart failure. Parenteral antibiotic treatment, nasal oxygen and diuretics were administered, and his symptoms completely disappeared within 10 days.

Case 2

A 39-year-old female patient was admitted to our hospital with the complaints of dyspnea on effort, cough and sputum. Her PA chest X-ray revealed elevation of the left hemidiaphragm, shifting of the mediastinal structures to the left and smaller rib cage on the left (Figure 3). No breathing sound on the left hemithorax was detected by auscultation. Complete blood count and biochemical analysis were completely normal. Neither lung parenchyma nor any vascular structure could be visualized at the left side on the thorax CT examination, and the right lung and mediastinal structures were found to be shifted to the left (Figure 4). In fiberoptic bronchoscopy, although the left main bronchus was shown to divide into upper and lower lobe bronchi, they ended immediately as a bronchial stump. Magnetic resonance imaging (MRI) angiography

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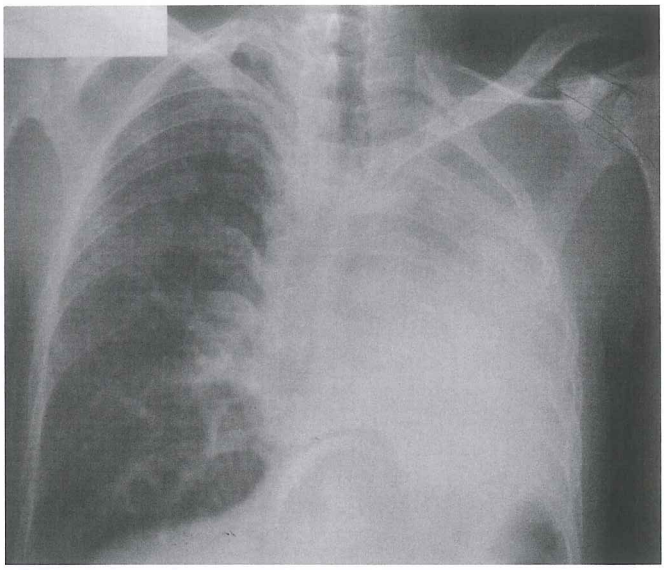

Figure 1. PA chest film of Case 1 showing compensatory hyperinflation of the right lung, mediastinal shifting to the left and total absence of the left lung.

revealed absence of the left pulmonary artery (Figure 5). The patient was diagnosed with left pulmonary agenesis, and her newly onset symptoms were attributed to acute bronchitis. The patient was placed on a symptomatic treatment and completely recovered after one week.

DISCUSSION

The incidence of pulmonary agenesis, aplasia and hypoplasia varies between 1/10,000 to 12,000 births [11]. Embryological basis of these pathological conditions is consistent with the hypothesis related to interference with normal development of the lungs between the 3rd and 24th

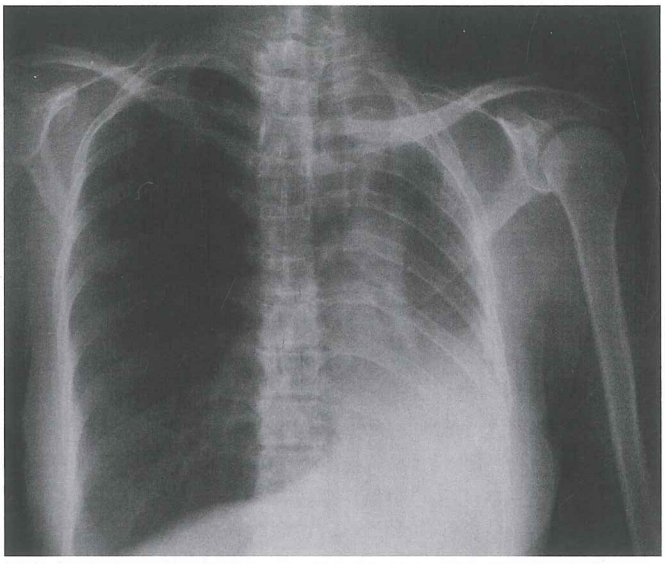

Figure 3. PA chest film of Case 2 showing compensatory hyperinflation of the right lung, herniation of the right lung parenchyma to the left, elevation of the left hemidiaphragm and shifting of the mediastinal structures to the left.

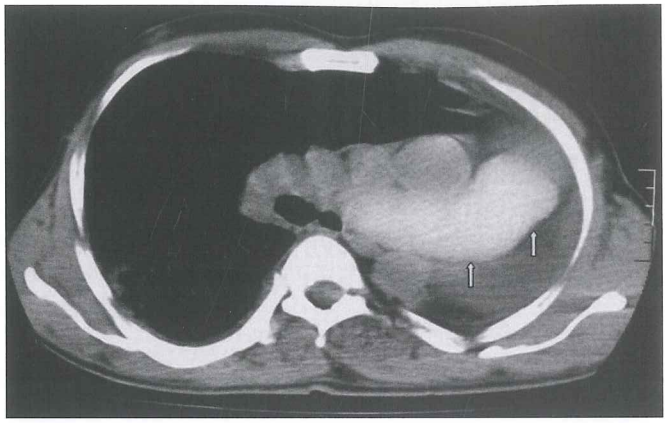

Figure 2. CT angiography of Case 1 showing herniation of the right lung to the left. The arrows indicate dilated main pulmonary artery and right pulmonary artery. The left pulmonary artery cannot be visualized.

weeks of gestation during the embryonic, pseudoglandular and canalicular periods [12]. Experimental evidence has implicated vitamin A deficiency, folic acid deficiency and salicylates in the causation of lung agenesis [5]. Clinical, radiological and pathological criteria permit the designation of a specific disease process in most cases; in some, however, it is impossible to categorize an anomaly precisely. Some features of the anomalies may be shared with multiple categories. For these reasons, agenesis and aplasia are usually considered together [11].

The clinical picture depends not only upon the degree of pulmonary abnormality, but also upon the presence of congenital malformations elsewhere. The most frequent concomitant extra-pulmonary abnormalities are those of chest wall, musculoskeletal system, ipsilateral face, diaphragm, abdominal wall, kidney and urinary tract [2,3]. These extra-pulmonary abnormalities also lend a strong support for the embryological basis of pulmonary agenesis-aplasia and hypoplasia. As presented here, Case 1 had abnormalities of the musculoskeletal system, ipsilateral face and kidney.

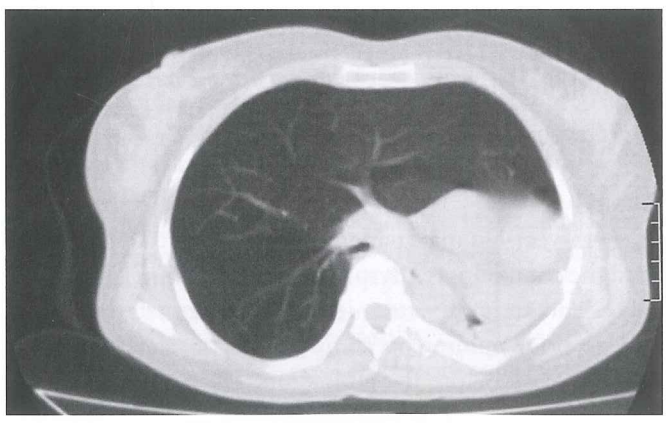

Figure 4. Thorax CT of Case 2 showing total absence of left lung and shifting of right lung and mediastinal structures to the left.

Figure 5. MRI angiography of Case 2: the arrow indicates the right pulmonary artery. The left pulmonary artery is totally absent.

Although the present evidence suggests that the patients with unilateral agenesis-aplasia usually die soon after birth [2-5], it was reported that some extreme cases survived into adulthood without any symptoms [8,9]. Our patients also had no pulmonary complaints before the index admissions. As was the case in the present report, such patients are recognized coincidentally in adulthood on the basis of abnormal chest roentgenograms taken for other reasons. Once they become symptomatic for any reason and are recognized, their differential diagnosis includes hyperlucent and hypoplastic lung syndromes and obstructive lung lesions, mainly lung cancer [11]. The preliminary diagnosis in our patients was also lung cancer.

The most frequent presentations on PA chest films are opacification and decreased size of the affected hemithorax, compensatory hyperinflation of the contralateral hemithorax, elevation of ipsilateral hemidiaphragm, absent ipsilateral and enlarged contralateral pulmonary artery shadow and ipsilateral shift of the mediastinum [2-5]. Our patients also demonstrated all of these findings (Figures 1,3). Recent reports strongly suggest that today's technological developments in radiology have obviated the need for invasive procedures, like the classical pulmonary angiography; definitive diagnosis can be easily made by MRI angiography or contrast-enhanced CT [6,7,13,14]. The final diagnosis in our cases was also made by the combination of contrast-enhanced CT and MRI angiographies.

We present these cases in view of their asymptomatic status until the fourth and fifth decades, a condition which is exceptionally rare, particularly in such major congenital pulmonary anomalies. We also stress that, although examples of most anomalies have been reported in children, respiratory physicians treating adults should also be aware of them.

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