Right Pulmonary Agenesis First Diagnosed in Adulthood

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Summary

Unilateral pulmonary agenesis is a rare condition and may be associated with some other abnormalities. Diagnosis is usually made in neonatal period or in early childhood. It is very rarely seen in adults. Patients who have no or mild associated anomalies may survive into adulthood. Since it is a rare condition we present an adult male case of right pulmonary agenesis who was diagnosed in adulthood.

Key Words: Agenesis, lung, congenital, anomaly

Özet

İlk Olarak Erişkin Yaşta Tanı Konmuş Sağ Akciğer Agenesisi (Olgu Sunumu)

Anahtar Kelimeler: Agenezi, akciğer, konjenital, anomali

Introduction

Unilateral pulmonary agenesis (UPA) is a rare condition and associated with some other abnormalities in more than 50% of the patients mainly involving the cardiovascular, gastro-intestinal, musculoskeletal, urogenital systems. UPA is usually seen in infancy or early childhood. It is not common in adults. Patients who have no or mild associated anomalies may survive into adulthood (1,2,3,4,5). Since it is a rare condition we present a case of right pulmonary agenesis who was diagnosed in our hospital.

Clinical Presentation

The patient was a 20 years old male who had complaints of dyspnea since childhood and intermittent cough for 6 years. Dyspnea complaint was continous and worsening during physical activities. His early diagnosis in childhood was dextrocardia and bronchitis. After conscripting the army he was referred to cardiology department of our hospital due to suspicion of dextrocardia. After cardiac evaluation, no cardiac abnormality was found and the patient was referred to our department. His physical examination revealed decreased breath sounds at auscultation on lateral and back side of right hemithorax and trachea was deviated to right side on palpation. Other sytmes were found as normal. Routine blood chemistry and complete blood count were in normal range except a low total cholesterol 131 mg/dL and a high LDH 336 U/L. Eritrocyte sedimentation rate was 2 mm/h. On chest X-ray; neither right hemidiaphragm could be discriminated nor a cardiac silhouette was detected but an almost homogeneous density was revealed in middle and lower right lung fields in right hemithorax. trachea showed right sided deviation and inter costal distances were increased and the ribs became parallel to each other in left hemithorax (Figure-1).
Contrast-enhanced computed tomography of thorax revealed that heart displaced to right and rotated. Left lung herniated into right hemithorax, expanded and hyperinflated (Figure 2). Pulmonary IV digital substracting angiography (DSA) examination revealed that right pulmonary vasculature could not be seen in right hemithorax and left pulmonary vasculature was normal (Figure 3). Magnetic resonans imaging (MRI) examination revealed that thoracic volume was reduced on the right side, trachea, heart and mediastinum displaced into right hemithorax and right diaphragm was markedly elevated and herniated into right hemithorax and cardiac chambers, ventricular walls and pericardium was normal (Figure 4). Fiberoptic bronchoscopy (FOB) revealed that trachea was showing a continuation with left main bronchus, there was no main carina and segment openings deviated postero-laterally.

Spirometric values were compatible with restrictive lung disease; FVC: 53% pred, FEV1: 51%pred, and FEV1/FVC: 95%pred. Final diagnosis was right pulmonary agenesis.

**Discussion**

Pulmonary agenesis means complete absence of carina, main bronchus, lung tissue and pulmonary vasculature while in pulmonary aplasia there is a carina and rudimentary main bronchus with absence of lung tissue and pulmonary vessels. Bilateral pulmonary agenesis extremely rare and incompatible with surviving. In practice an etiological, pathogenetic or clinical distinction between agenesis and aplasia is rare and the two conditions are usually considered together (5). Its frequency is estimated at 1:15,000 autopsies (2). Aetiology is not completely known but vitamin A deficiency, viral agents, or genetic factors are discussed.
More than 50% of patients with unilateral agenesis have other associated anomalies mainly involving the cardiovascular, gastro-intestinal, musculoskeletal and urogenital systems (1,4,5). Our case did not have any associated anomaly. UPA results from an insult during the fourth week of fetal life. It was found that all cases of pulmonary hypoplasia/agenesis associated with radial ray defects or hemifacial microsomia have ipsilateral malformations. This suggests a vascular etiology due to the nature of the paired dorsal aortic arches at this stage of development(6). Pulmonary agenesis occurs on the right sight more than the left side (1). Right pulmonary agenesis is more frequently associated with congenital malformations of other systems than left pulmonary agenesis(11).

Diagnosis is usually made in neonatal period or in early childhood (1,2,6). There are some cases diagnosed antenatally (2,8,9). Chest X-ray is the key examination which lead to physician further examination. Chest X-ray usually reveals mediastinal and tracheal shift ipsilaterally but exceptions may occur (4). It also shows narrowing of ribs ipsilaterally and broadening of them contralaterally with hyperinflation. It may be hard to determine a cardiac silhouette and diaphragmatic contour. All were valid for our case. Contrast enhanced computed tomography is almost definitive for the diagnosis (10). It may show main vascular and bronchial structures as well as lung parenchyma. It also helps to determine existence of carina to distinguish agenesis from aplasia. Angiography or DSA might be necessary to show the vasculature. Angiography is a more accurate but a more invasive method than DSA. In our case, with respect to less invasiveness of the method, DSA was used. MRI and/or magnetic resonance angiography techniques may be used for the diagnosis (8) and only MRI methods might be enough for diagnosis of UPA. In our case MRI definitively showed existence of UPA.

FOB is very helpful to observe the bronchial tree as well as the absence or existence of carina and a rudimentary main bronchus to discriminate agenesis from aplasia. Our case was diagnosed with contrast enhanced CT, DSA, MRI and FOB. Another case of left pulmonary aplasia from another military hospital (10) the definitive diagnosis was made with contrast enhanced tomography and MR angiography without bronchoscopy. Clinical presentation may vary depends on associated anomalies and/or existence of abnormalities in healthy lungs such as bronchiectasis. Some patients, those with no associated anomalies and/or no bronchiectasis or no recurrent respiratory infections might be symptom free or with minimal symptoms (4). These patients may form the majority of the patients who survive into young adulthood or adulthood which is not common. Patients who have symptoms could be misdiagnosed as bronchitis, recurrent pulmonary infections or cardiac disorders. Our patient was diagnosed as bronchitis and dextrocardia before referring to our hospital. Dyspnea and tachycardia are the most frequent symptoms. Stridor, wheezing and coughing are other frequent symptoms (3). Compatible with this, our patient had dyspnea and cough. This may result from limited lung area and different lung kinetics due to abnormal bronchial tree anatomy. On the other hand these patients may have recurrent lung infections which is mainly due to bronchiectasis, rudimentary bronchus in aplasia or abnormal lung kinetics. In our country adult males might have greater chance to be diagnosed than adult females because Turkish Armed Forces has a well working health system with periodical screening of every soldier. Our case was an example of this.

Differential diagnosis includes total atelectasis, pneumonectomy, pulmonary hypoplasia, diaphragmatic evantration, pneumonia and pleural effusion (3).

We conclude that UPA should bear in mind when a physician confronted with a chest-X ray compatible with marked mediastinal displacement in an adult even though it is a rare condition.

References