Right-side pulmonary agenesis with atrial septal defect in adult

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Abstract
Pulmonary agenesis, a rare congenital condition, is incompatible with life when present bilateral, while unilateral agenesis is usually detected in infancy or early childhood. Rare asymptomatic patients may reach adulthood undiagnosed, with signs mimicking common conditions presenting as radiopaque hemithorax with ipsilateral mediastinal shift. Here, we describe a case of a young lady, with history of consanguinity, who presented with complaints, suggestive of lower respiratory tract infection, and was investigated and diagnosed to be a case of right-side pulmonary agenesis with large ostium secondum atrial septal defect. Our present case emphasizes the importance of presence of pulmonary agenesis with cardiac congenital anomaly, remaining asymptomatic until adulthood, particularly in patients born of parents with consanguineous marriages.

Introduction
Pulmonary agenesis could be defined as non-development of pulmonary vessels, bronchi, and lung parenchyma. It is an extremely rare disorder, resulting from failure of development of embryonic lung bud. Its prevalence varies from about 34 to 100 per million live births [1]. About 70% cases occur on the left side, and there is no gender predisposition. Unilateral lung agenesis was first reported in 1673 by De Pozze, as autopsy finding in a female adult. This congenital defect may be found alone or may be associated with anomalies of other organ systems, in about half of the cases, involving cardiovascular, musculoskeletal, gastrointestinal, or genito-urinary systems. Consanguinity has been reported in patients with pulmonary agenesis with autosomal recessive mode of inheritance [2]. Presence of this anomaly generally comes to light in infancy because of recurrent chest infections, or because of cardiovascular insufficiency, but rare cases may survive up to adulthood, without many health problems. The oldest patient reported was 72 years old. We hereby present a rare case of a patient with right-side congenital pulmonary agenesis, along with large ostium secondum type of atrial septal defect, born of parents with consanguineous marriage, surviving up to adulthood without significant health problems.

Case Report
A young lady, aged 25 years, with history of consanguinity, presented with complaints of breathlessness on exertion, low-grade fever, and a mild dull aching type of right-sided chest pain for the past 10 days. On clinical examination, she was febrile and looking sick. Chest examination revealed decreased breathing movements and overcrowding of ribs on right side; trachea along with apical cardiac impulse was shifted to right. We observed impaired note on percussion and decreased breath sounds, with no adventitious sounds on right side.

Her chest radiograph postero-anterior view (Fig. 1) revealed homogenous radiopacity and signs of volume reduction on right side, including overcrowding of right-sided ribs, pulled-up right hemidiaphragm, and scoliosis with...
concavity on right side. Her sputum smear examination for acid-fast bacilli, of morning mucoid sample, from Revised National Tuberculosis Control Program laboratory, was negative. Her spirometric analysis revealed a restrictive ventilatory defect. Provisional diagnosis of right-sided pulmonary agenesis was made. She was advised echocardiography to rule out presence of any congenital cardiac anomaly, which revealed large ostium secundum type of atrial septal defect, with left to right shunt, and moderate tricuspid regurgitation. No pulmonary arterial hypertension was found. Right pulmonary artery was rudimentary. Her ultrasonography of abdomen was normal, and without any congenital anomaly. She was further subjected to computerized tomographic scan of thorax (Fig. 2), which reported non-visualized right lung, with complete shift of mediastinum to right side, along with marked crowding of ribs and loss of volume of right hemithorax. With this lead on investigation, patient underwent bronchoscopy, which revealed shift of trachea and carina to right and a small pit at the site of right main bronchus.

A diagnosis of right-sided unilateral pulmonary agenesis with large ostium secundum type of atrial septal defect was made. Patient was given antibiotics of non-tubercular potential, to clear her lung infection, to which she responded satisfactorily, and was discharged after a hospital stay of 7 days, with advice to attend cardiovascular thoracic surgery unit, for surgical management of atrial septal defect.

**Discussion**

The most common radiological picture of unilateral pulmonary agenesis is unilateral thoracic opacification and decreased size of affected side of hemithorax, compensatory hyperinflation of opposite lung, and ipsilateral shift of mediastinum [3]. Our patient had all these features, with ipsilateral scoliosis, in addition.

The embryological origin of lung agenesis is estimated to be around 4 weeks of gestational age. It results from failure of primitive lung bud or bronchial bud to develop properly. Spencer modified earlier classification and divided pulmonary agenesis into three groups: (1) bilateral complete agenesis; (2) unilateral agenesis; and (3) lobar agenesis or lesser forms of congenital anomaly. Etiology of pulmonary agenesis is largely unknown, although viral, genetic factors, chromosomal anomaly, salicylates, and folic acid or vitamin A deficiency are thought to be involved.

Because of its complications, it is generally diagnosed soon after birth, but rarely patients may survive up to adulthood, without much complication. Only few adult presentations have been reported in the literature and that is also the case with those patients who do not have associated congenital anomalies of other organ systems [4]. Right-side pulmonary agenesis carries the poorest prognosis and highest mortality risk, because of its association with congenital anomalies of...
other organ systems [5]. Prognosis is better if there is unilateral left-side pulmonary agenesis, without any associated cardiovascular malformations [5]. About 50% of patients with pulmonary agenesis have other associated anomalies including patent ductus arteriosus, ventricular septal defect, atrial septal defect, narrow trachea, and tracheo-esophageal fistula.

Our patient is a rare case, because she has survived up to adulthood, without many health problems, despite unilateral right-side pulmonary agenesis and large ostium secondum type of atrial septal defect. Differential diagnosis of this patient included complete resorption or cicatrisation atelectasis, pneumonia, pleural effusion, thickened pleura, pneumonectomy, lung hypoplasia, and complete agenesis of right lung.

Present case has highlighted the clinical fact that pulmonary agenesis could be a rare but an important cause for clinical and radiological features suggestive of unilateral volume loss, particularly in patients with history of parental consanguinity.

**Disclosure Statements**

No conflict of interest declared. Appropriate written informed consent was obtained for publication of this case report and accompanying images.

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