The eldest survivor in Malaysia: Pulmonary agenesis with isolated dextrocardia in late adulthood

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SUMMARY

Pulmonary agenesis is a rare congenital anomaly. It is frequently associated with other congenital anomalies and is present during early childhood. The exact aetiology of pulmonary agenesis is not fully understood. In this case report, we present a rare case of type 1 right pulmonary agenesis with isolated dextrocardia in an elderly man, diagnosed at 68 years old. He was well antenatally with no previous history of hospitalisation or respiratory-related health issues. Upon his short presentation with upper respiratory tract symptoms, a chest radiograph performed shows unilateral right hemithorax opacity with crowding of the right ribs. Subsequent contrast-enhanced computed tomography (CT) of the thorax confirmed features of right pulmonary agenesis with isolated dextrocardia. Despite having the type of congenital pulmonary underdevelopment with the worst prognosis, this man has survived into his late adulthood without any significant symptoms. To the author's knowledge, diagnosing pulmonary agenesis with isolated dextrocardia at this age is the oldest age that has been reported in Malaysia. This case highlights features of pulmonary agenesis as well as the importance of considering this rare condition as a differential diagnosis for unilateral hemithorax opacity in adult chest radiographs. The advancement of CT nowadays has allowed us to utilise various imaging techniques to avoid invasive methods such as bronchoscopy, bronchography, or angiography to establish this diagnosis.

INTRODUCTION

Pulmonary agenesis is a rare congenital anomaly with a prevalence of 24-34 out of 1,000,000 live births and a slight female preponderance.[1] It is a condition where there is unilateral absence of developed pulmonary vessels, bronchi, and parenchyma.[2] This diagnosis is usually made during childhood, with approximately a fifty percent survival rate due to concomitant anomalies.[3] The incidence of right and left lung pulmonary agenesis is equal in reported cases, but right pulmonary agenesis manifests a worse overall prognosis compared to left pulmonary agenesis.[4]

The first description of this condition was narrated back in 1673 by De Pozze as an incidental finding during the autopsy of an adult female.[5] As this is a congenital anomaly, most of the cases reported are diagnosed within the paediatric population, as these patients are usually symptomatic either from the pulmonary agenesis itself or other concomitant anomalies. To date, the oldest age reported for the first diagnosis of pulmonary agenesis in Malaysia is 40 years old, as per Sulaiman et al., in their article published in June 2020.[6]

In this case report, we present a case of type 1 right pulmonary agenesis with isolated dextrocardia, first diagnosed at the age of 68 years old, which is the oldest reported age in Malaysia.

CASE PRESENTATION

Mr. R, a 68-year-old man, is a retired farmer who has lived in a small village his entire life. He has underlying type 2 diabetes mellitus and hypertension, which are well controlled with oral medication under community health clinic follow-up. He is also a chronic smoker with 12 pack-years. There was no previous history of surgery or hospitalisation.

Mr. R presented to the emergency department in July 2017 for a short two-day history of dry cough and shortness of breath. He has no documented fever. Upon examination, he was not tachypnoeic or appeared to be in distress. He was well perfused with a capillary return time of less than 2 seconds. His vital signs were all within normal limits, with a heart rate of 70 bpm, a respiratory rate of 18, and oxygen saturation of 97% under room air. However, on auscultation, there was reduced right lung breath sound with rhonchi on the left lung. Otherwise, his laboratory investigations, namely the full blood count, renal profile, and electrolytes, were normal.

An erect frontal chest radiography was performed in view of reduced right lung breath sounds on auscultation. Surprisingly, his chest radiograph (Fig. 1) shows unilateral right hemithorax opacity with crowding of the ribs, which did not fit into his relatively stable presentation. The tracheal and mediastinal were deviated to the right in keeping with right lung volume loss. The right main bronchus was not well visualised in this radiograph. On the other hand, the left main bronchus was patent, and the left lung field was clear. There was an absence of a left cardiac silhouette on his radiograph. An additional remark on this radiograph is that the stomach bubble was seen in a normal position, inferior to the left hemidiaphragm.

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Table I: Differential diagnosis to be considered for complete opaque hemithorax.

Non-displaced mediastinum	Displaced mediastinum towards opacity	Displaced mediastinum away from opacity
• Consolidation • Pleural effusion • Chest wall/pleural mass	• Total lung collapse • Pneumonectomy • Pulmonary hypoplasia/agenesis • Malignancy	• Large pleural effusion • Large pulmonary mass • Diaphragmatic hernia

An initial diagnosis of bronchiolitis was made, and Mr. R responded well to salbutamol nebulisation. He was subsequently discharged from the emergency department with prophylactic antibiotics. An outpatient computed tomography (CT) thorax and respiratory clinic referral was made for further investigation.

The contrast-enhanced CT thorax (Fig. 2) shows the absence of the right lung parenchyma, right main bronchus, and right pulmonary vessels. The heart was situated on the right side of the thorax with its apex pointing to the right, suggestive of dextrocardia. The ascending aorta was seen arising from the left ventricle and coursing to the left to resume normal position of the descending aorta. The trachea and oesophagus were intact but deviated to the right. There was hyperplasia of the left lung with a normal left main bronchus and pulmonary vessels. No focal lung nodule or lesion noted within. Other negative findings include no pleural effusion, bronchiectasis, or mediastinal lymphadenopathy. The visualised upper abdominal organs were in normal position in keeping with situs solitus.

To further visualise the structures of the heart and great vessels, we have reconstructed volume rendering images to provide a 3-dimensional visualisation, as seen in Figure 3. This volume rendered images help to ascertain the absence of the right pulmonary vessel, which are the main differentiating features of pulmonary agenesis. Overall features of this CT thorax are suggestive of type 1 pulmonary agenesis with isolated dextrocardia. Fortunately, Mr. R was discharged well and does not require any long-term follow-up or further intervention.

DISCUSSION

Pulmonary agenesis is a rare congenital anomaly that is usually present in childhood. Uncommonly but possible, patients may present late in adulthood without developing significant symptoms to seek medical treatment earlier in life, such as in this case where Mr. R has survived with one lung until his late adulthood.

The exact aetiology of pulmonary agenesis is not fully understood. It has been postulated to occur due to arrested development of the lung during the 4th gestational week, where the embryogenesis of the pulmonary system begins via formation of the respiratory diverticulum from the laryngotracheal bud. The intrauterine development of the lung has been divided into five phases throughout the entire pregnancy, namely the embryonic, pseudoglandular, canalicular, saccular, and alveolar phases.[7] Some hypothesised that during the 4th week of gestation, abnormal blood flow in the dorsal aortic arch causes pulmonary agenesis.[5] Genetic factors, viral agents, and dietary deficiency of vitamin A or folic acid or maternal use of salicylates have been suggested as contributing factors.[7]

Pulmonary agenesis is rare, and therefore recognising its radiological features will be challenging, particularly when it's not in the common age group. Understanding the differential diagnosis for unilateral complete opaque hemithorax is essential when it comes to chest radiographs. According to Chapman & Nakielny's Aids to Radiological Differential Diagnosis,[8] the differential diagnosis of complete opacity of unilateral hemithorax can be further narrowed depending on the mediastinal displacement, as stated in Table I.

In our case, Mr. R's chest radiograph showed right hemithorax opacity with displaced mediastinum towards opacity, which narrowed down the differential to total lung collapse and pulmonary agenesis, as he had no previous surgery.

Prenatal diagnosis of pulmonary hypoplasia or agenesis is usually done by ultrasound or magnetic resonance imaging (MRI) to reduce radiation dose to the foetus.[9] On the other hand, diagnosing pulmonary hypoplasia or agenesis in adults can be achieved effortlessly via contrast-enhanced CT thorax. The advancement of CT nowadays allows not only soft tissue and lung window views, but also reconstruction of volume rendering images to better appreciate the great vessels and airways. Hence, there is no need for more invasive pulmonary angiography, bronchoscopy, or bronchography to delineate the pulmonary vessels and bronchus pattern.

Pulmonary hypoplasia has been classified into three types by Boyden, which was then modified by Schneider in 1912, as stated below.[2]

- Type 1 (Agenesis): Complete absence of lung and bronchus with no vascular supply to the affected side.
- Type 2 (Aplasia): Rudimentary bronchus with complete absence of pulmonary parenchyma.
- Type 3 (Hypoplasia): Presence of variable amounts of bronchial tree, pulmonary parenchyma, and supporting vasculature.

In our case, Mr. R's CT showed complete absence of right lung parenchyma and right pulmonary vessels with no rudimentary bronchus, in keeping with type 1 pulmonary agenesis.

Pulmonary agenesis is often associated with a wide range of congenital anomalies in half of the affected individuals. For instance, the cardiovascular (patent ductus arteriosus, patent foramen ovale), gastrointestinal (tracheoesophageal fistula, imperforate anus), genitourinary, or musculoskeletal system (limb or vertebral anomalies). Depending on the severity of

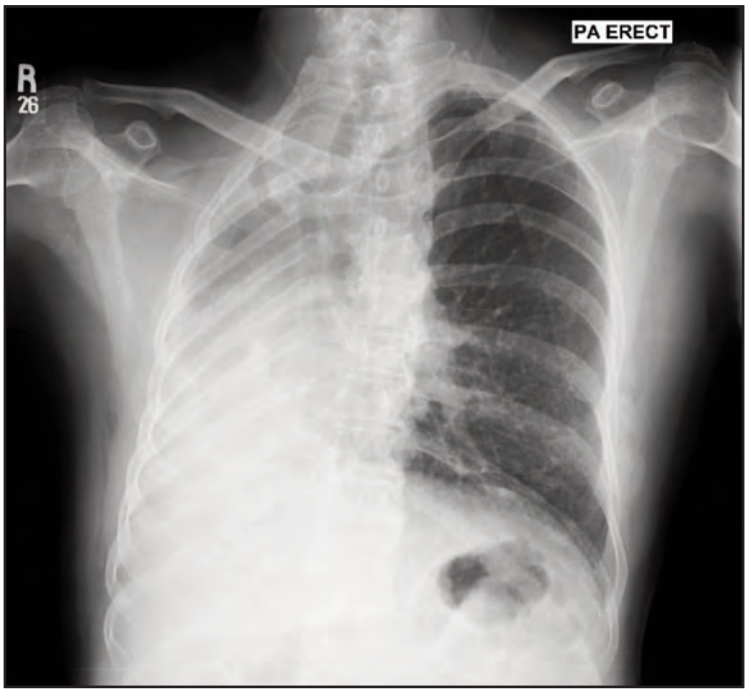

Fig. 1: Erect frontal chest radiograph. Complete opaque right hemithorax with crowding of right ribs. Tracheal and mediastinal shift to the right, resulting in the absence of normal left cardiac silhouette. Normal stomach gas bubble position in the left hypochondriac.

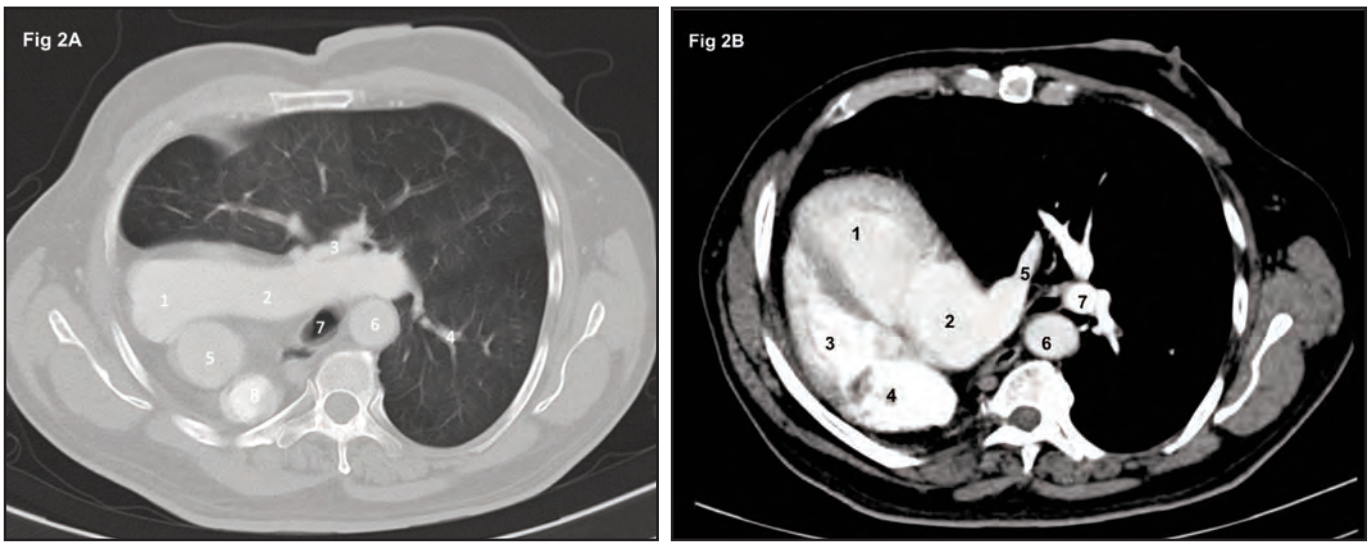

Fig. 2: A) Axial image of CECT Thorax at the level of mid thorax showed mediastinum deviated to the right hemithorax with absent right lung and hyperplasia of the left lung. 1. Pulmonary trunk, 2. Left main pulmonary artery, 3. Left upper lobe pulmonary artery, 4. Left lower lobe pulmonary artery, 5. Ascending aorta, 6. Descending aorta, 7. Trachea, 8. Superior vena cava, B) Reconstructed axial image of CECT Thorax at the level of the heart showed the apex pointing to the right in keeping with dextrocardia. 1. Left ventricle, 2. Left atrium, 3. Right ventricle, 4. Right atrium, 5. Left inferior pulmonary vein, 6. Descending thoracic aorta, 7. Left segmental pulmonary artery.

the accompanying comorbid anomaly, variable clinical findings can occur. Fortunately, Mr. R does not possess any of these congenital concomitants, which is probably the reason he survived asymptomatically until his late adulthood. There is no definite treatment for pulmonary underdevelopment to the current date, and the management is mainly supportive therapy.

CONCLUSION

This case is a type 1 pulmonary agenesis with isolated dextrocardia reported and diagnosed at the oldest age in Malaysia, discovered during a work-up for bronchitis. This case is a rare presentation that reminds all clinicians to include this differential diagnosis in their minds in the future. Recognising pulmonary agenesis in adult patients can be challenging with chest radiographs alone, but the current

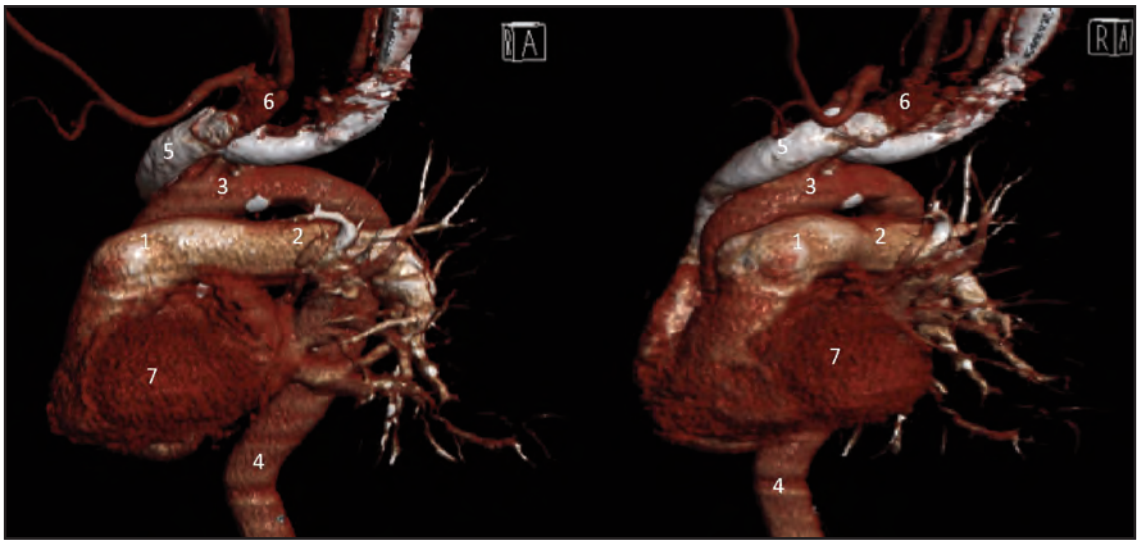

Fig. 3: Reconstructed volume rendering images of the heart and great vessels showing the absence of the right main pulmonary artery. Annotation: 1. Pulmonary trunk, 2. Left main pulmonary artery, 3. Arch of the aorta, 4. Descending thoracic aorta, 5. Superior vena cava, 6. Brachiocephalic trunk, 7. Heart (right ventricle).

advanced CT imaging tools, such as volume-rendered images, will aid in the diagnosis of this condition. It is not necessary to further investigate with more invasive methods such as bronchoscopy, bronchography, or angiography to establish this diagnosis.

DECLARATION

The authors declare no actual or potential conflict of interest in relation to this article.

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