Radiology Case Reports 18 (2023) 3724–3728

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journal homepage: www.elsevier.com/locate/radcr

Case Report

Incidental pulmonary agenesis with multiple associated anomalies: A case report ☆

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ARTICLE INFO

Article history:
Received 15 March 2023
Revised 29 July 2023
Accepted 1 August 2023

Keywords:
Pulmonary agenesis
Unilateral
Congenital
Pulmonary underdevelopment

ABSTRACT

Pulmonary agenesis is an uncommon congenital abnormality of the lungs that occurs either in isolation or in association with other anomalies in several organ systems. Right sided pulmonary agenesis is reported to have a worse prognosis owing to the more severe mediastinal shift and resultant compression of mediastinal structures. We present a case of right-side pulmonary agenesis with multiple associated anomalies found incidentally in a 19-year-old female patient.

Introduction

Pulmonary agenesis is encompassed under the umbrella term pulmonary underdevelopment or lung-agenesis-hypoplasia complex [1,2]. It is a rare congenital anomaly which occurs with an incidence of 1 to 2 cases per 10,000 live births. Bilateral pulmonary agenesis is extremely rare and incompatible with life [3]. No side predilection is noted in unilateral pulmonary agenesis, but patients with right lung agenesis have a shorter life span owing to a more significant shift and compression of vital mediastinal structures [1,2]. It can exist in isolation, but associated malformations occur in more than 50% of the cases, including anomalies of the cardiovascular, skeletal, gastrointestinal and genitourinary systems [1]. In 1988, Knowels et al. [4] suggested that pulmonary agenesis may be seen in association with the VACTERL (vertebral anomalies, anorectal anomalies, cardiac anomalies, trachea-esophageal fistula, renal anomalies and limb anomalies) sequence as an alternative to trachea-esophageal fistula after making note of several such cases. Clinical presentation is quite variable and ranges from severe early respiratory distress after birth to incidental detection of the anomaly in an asymptomatic adult [1,3].

Imaging is usually required for diagnosis of pulmonary agenesis, with chest radiography often being the first modality to bring this condition into consideration. An opaque hemithorax with ipsilateral mediastinal shift is the typical radiographic feature. The diagnosis can be ascertained by further imaging studies including CT, MRI, bronchography and angiography [1]. Cross sectional imaging modalities have the

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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https://doi.org/10.1016/j.radcr.2023.08.002

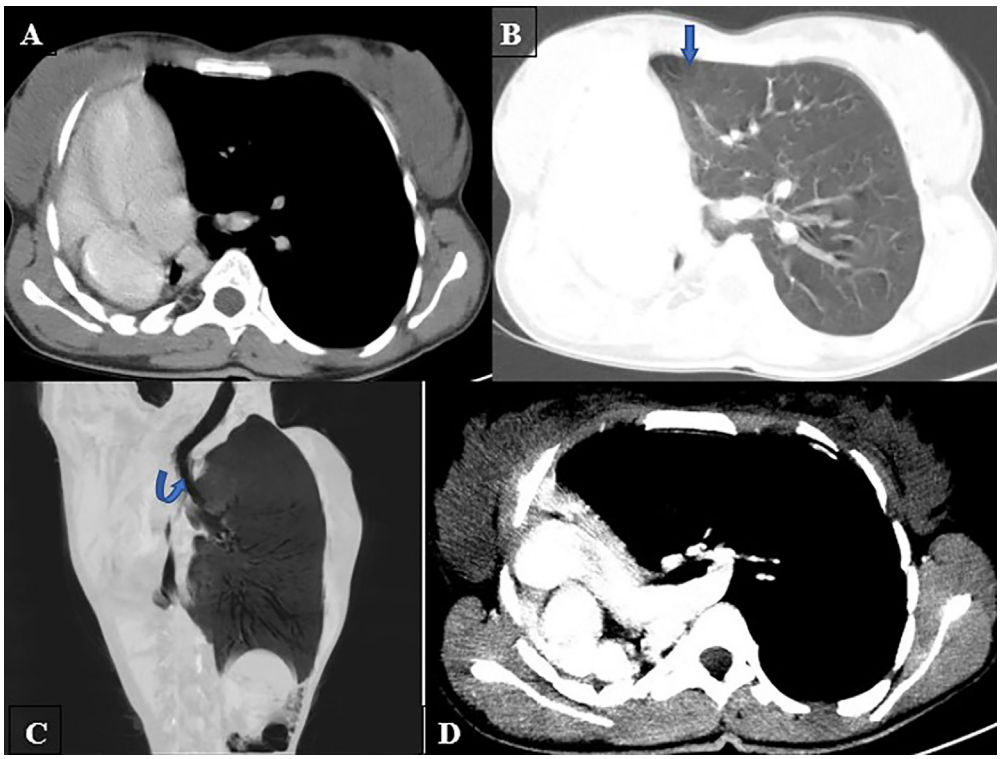

Fig. 1 – Axial CT-Chest in mediastinal (A) and lung window (B) settings show complete absence of the right lung with the heart occupying the right hemithorax. Compensatory hyperinflation of the left lung with anterior herniation across the midline (arrow in B) is also seen. Oblique coronal MinIP image (C) shows the total absence of a right bronchial bud with the trachea continuing as the left main bronchus (curved arrow in C). Axial MIP (D) at the level of the pulmonary trunk shows absence of the right pulmonary artery.

added benefit of assessing for associated anomalies in other body systems.

Case report

We present a case of a 19-year-old previously well female adolescent who presented with signs of upper respiratory tract infection and dry cough of a few days duration. As part of the initial work up, a chest radiograph was obtained which showed an opaque right hemithorax with marked rightward mediastinal shift. This finding prompted acquisition of the CT chest presented here (Fig. 1) which demonstrated complete absence of the right lung, right mainstem bronchus and right pulmonary artery. Compensatory hyperinflation of the left lung with herniation across the midline was also noted.

The patient also had fusion of the vertebral bodies and posterior elements of multiple lower cervical and upper thoracic vertebrae (Fig. 2), which is diagnostic of Klippel-Feil syndrome.

Bilateral cervical ribs arising from the C7 vertebrae were also seen (Fig. 3).

In the upper abdomen (Fig. 4), the left kidney was not visualized in its expected anatomic location. Abdominal ultrasound was subsequently done (not shown), the left kidney was not visualized elsewhere in the abdomen or pelvis. Echocardiography done to look for associated cardiac anomalies was negative (not shown).

Conservative management with occasional follow-up was recommended, as the patient was doing well. She quickly recovered from her upper respiratory tract infection and appeared to be doing well on subsequent visits at the outpatient clinic.

Discussion

Pulmonary underdevelopment was initially classified in 1912 by Schneider and Schwalbe and modified by Boyden in 1955 into 3 groups [1,5]. This classification is based on the stage of development of the primitive lung bud. Group I, also called pulmonary agenesis is characterized by the complete absence of unilateral lung parenchyma, bronchus, and vasculature. In group II, namely pulmonary aplasia, there is a complete absence of unilateral lung with a rudimentary blind-ending bronchus. In group III, pulmonary hypoplasia, there is bronchial hypoplasia with a variable amount of remaining lung tissue. Our patient had type I disease.

The exact etiology of pulmonary agenesis is unknown but it has been hypothesized that abnormal blood flow in the dorsal aortic arch during the fourth week of gestation causes

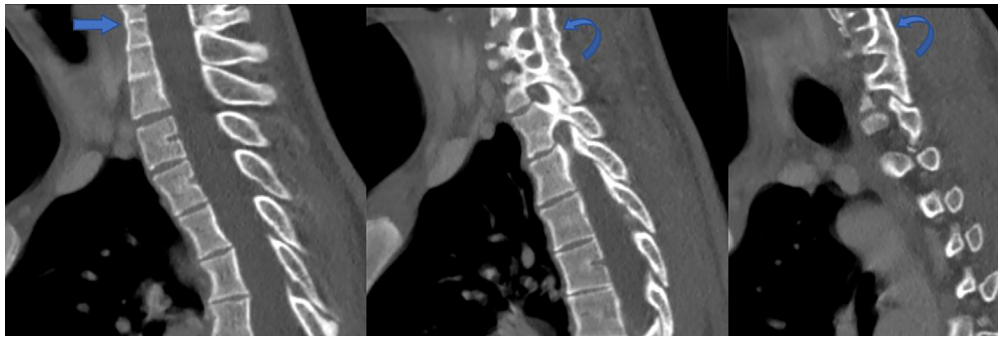

Fig. 2 – Sagittal bone window CT images at different levels demonstrate fusion of multiple lower cervical and upper thoracic vertebrae (C5-T1). Fusion of the vertebral bodies with the wasp-waist sign (arrow) is shown at the C5/C6 level. There is also partial fusion of the posterior elements of the mentioned segments (curved arrows).

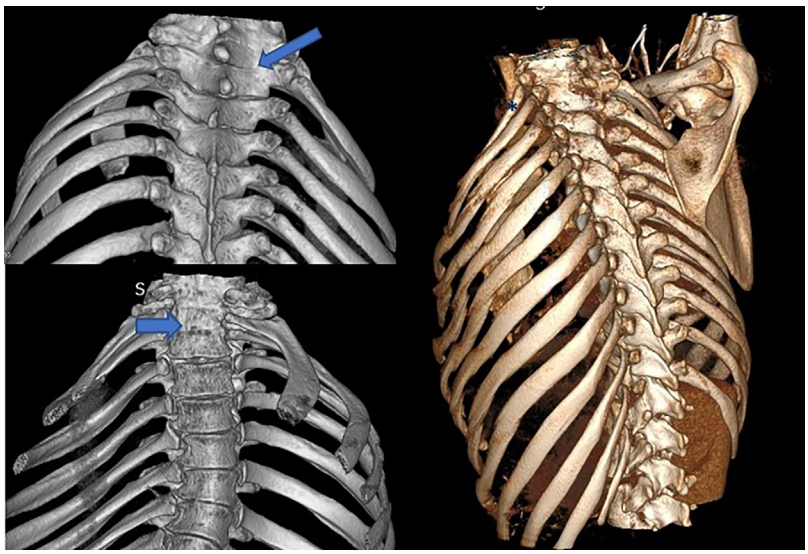

Fig. 3 – Bone algorithm 3D images better demonstrate the fusion of the lower cervical vertebral bodies and posterior elements (arrows). Bilateral cervical ribs arising from the C7 vertebrae (asterisk) are also noted.

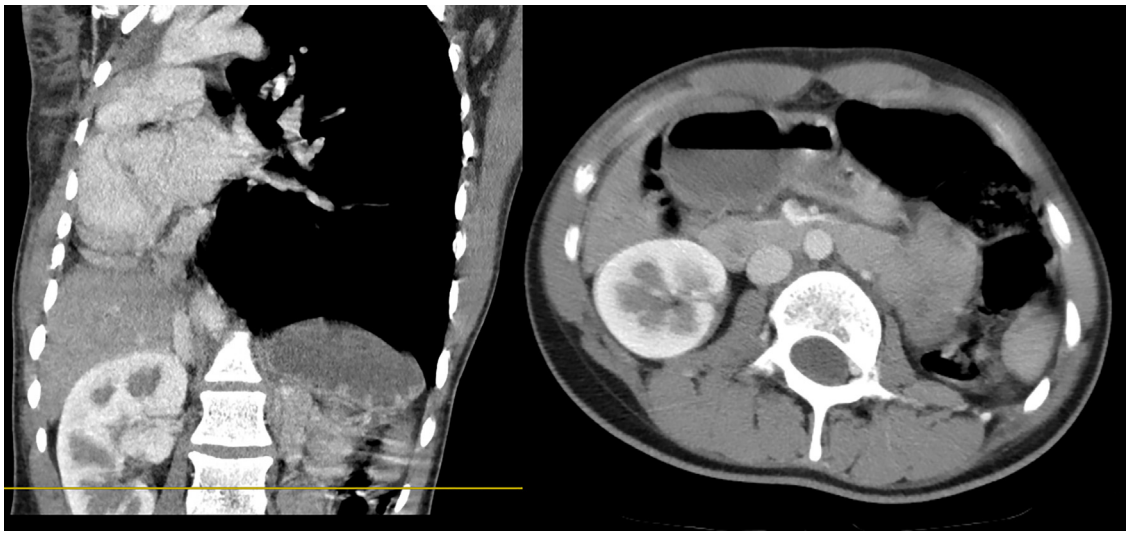

Fig. 4 – Coronal and axial CT images through the included upper abdomen shows the absence of the left kidney in its normal anatomic location. The left kidney could not be found elsewhere in the abdomen or pelvis on sonography (not shown).

pulmonary agenesis. Other possible causes included genetic, teratogenic, and mechanical factors [1].

The abnormality is usually unilateral, with no evidence of any side or gender predilection. More than half of cases have associated congenital anomalies that involve the cardiovascular, gastrointestinal, skeletal, and genitourinary systems [6]. Most limb and spinal malformations are ipsilateral to the pulmonary agenesis. The contralateral lung demonstrates compensatory physiologic hypertrophy but is otherwise normal [1].

Due to advancements in ultrasound and routine prenatal care, the condition can be diagnosed antenatally using ultrasound which shows a hyperechoic hemithorax. This can be confirmed with fetal MRI [7]. But most of the time, the diagnosis of pulmonary agenesis is first suspected at chest radiography which demonstrates a completely opaque hemithorax with rib crowding and marked mediastinal shift to the ipsilateral side such as in our patient. A normal contralateral lung with compensatory hyperinflation and associated herniation across the midline is a usual finding [8].

Abnormal segmentation of the vertebral bodies may be present as well, such as in our patient who was found to have fusion of the vertebral bodies and posterior elements of multiple lower cervical and upper thoracic vertebrae which is diagnostic of Klippel- Feil syndrome. Our patient also had bilateral cervical ribs. The literature shows that most rib anomalies are varied and can be found on either side with no impact on prognosis or management found in literature [1].

Cross-sectional imaging like CT and MRI provide important diagnostic information, confirming the absence of lung parenchyma, bronchial tree, and pulmonary vessels on the affected side and showing any associated anomalies if present [1]. Bronchoscopy can be done to verify that the mainstem bronchus is completely missing, and angiography demonstrates the absence of pulmonary and bronchial arteries on the side of the absent lung. Nowadays, these modalities are rarely necessary as cross-sectional imaging alone can provide exquisite anatomical details. Early detection is also necessary to reduce the development of fibrosis in patient's solitary lung which can be a sequelae of recurrent chest infections [10,11].

The prognosis varies and depends on the function of the existing lung and associated anomalies particularly severe heart and vessel malformations [6,9,10]. The mortality is reported as 33% during the first year of life and 50% in the first 5 years of life [9,10]. Patients with pulmonary agenesis tend to have a shorter life expectancy, even in the absence of other associated anomalies with most patients reported in the literature not surviving into the second decade; however, the oldest reported survivor lived to age 72 [6].

There is no specific therapy for pulmonary agenesis with most in the literature recommending a case-by-case symptomatic approach [9]. Treatment options include both medical management and surgical repair. Medical treatments consist of control of recurrent chest infection, bronchodilators and pulmonary rehabilitation when necessary. Surgery is usually needed in the case of associated congenital anomalies [9–11]. For example, patients having stumps (hypoplastic bud) may require surgical repair if antibiotics fail to control recurrent infection [11].

In summary, we present a 19-year-old female patient with incidentally found right-side pulmonary agenesis with multiple associated anomalies. Conservative management with occasional follow-up was recommended, as the patient was doing well.

Learning points

- Complete pulmonary agenesis (with absence of contralateral bronchus and pulmonary vessels) is a rare congenital anomaly of the lungs.
- Associated anomalies in other organ systems are found in more than half of these patients.
- Cases of right-side pulmonary agenesis are said to have worse outcomes owing to significant mediastinal shift and compression of vital structures.

Patient consent

Written informed consent was obtained from the patient to publish this case report. Any personal details and diagnostic images were anonymized to meet the confidentiality requirements.

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