CASE REPORT

Unilateral pulmonary artery agenesis with vertebral anomaly

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SUMMARY

We report a two-and-half-year-old boy who presented with recurrent respiratory tract infections. He had cor triatrium of right atrium, spina bifida occulta, hemivertebra and dysplastic right thumb. On CT of chest, he had also unilateral pulmonary artery agenesis. The case is being reported because of common manifestations of rare disease and its associated cardiac and skeletal abnormalities.

BACKGROUND

Unilateral pulmonary artery agenesis (UPAA) is a rare congenital anomaly often associated with other cardiovascular abnormalities such as tetralogy of Fallot, septal defects, coarctation of the aorta, subvalvular aortic stenosis, transposition of the great arteries, pulmonary stenosis; Taussig-Bing malformation and scimitar syndrome.¹ Most patients of UPAA have coexisting cardiac defects and present in infancy or childhood with symptoms related to the associated cardiac abnormality. Although less frequent, UPAA also may occur as an isolated finding. Subjects with isolated unilateral absence of the pulmonary artery may be asymptomatic or have recurrent respiratory infections. We report a case of UPAA with cor triatrium of right atrium, spina bifida, hemivertebra and dysplastic right thumb, which is not reported till date.

CASE PRESENTATION

A two-and-half-year-old boy presented with a 15-day history of fever, cough and fast breathing. There was no history of cyanosis, seizures, altered sensorium and any septic foci. He was born at full term by spontaneous vaginal delivery and perinatal

period was uneventful. He had attained developmental milestones at an appropriate age. There was a history of recurrent such episodes since 1 month of age at an interval of 2–3 months, which lasted for 15 days. The child was examined by a local doctor and received oral and intravenous antibiotics. There was a history of hospitalisation for similar illness 2 months back.

On examination, he was tachypnoeic (RR: 62/min) with subcostal and intercostal retractions. Capillary refill time was 3 s and temperature was 102°F. Pulses were well palpable. There was no cyanosis. The child was malnourished and stunted (weight: 60% and height: 84% of expected of WHO growth chart). His right hand had dysplastic thumb and thenar atrophy (figure 1). There was hyperlaxity of right wrist joint.

Examination of chest showed fullness of the left side and drooping of right shoulder. Scoliosis was present with convexity towards left. On percussion, there was dull note on the right side and resonant on the left side. On auscultation, bronchial breath sounds were heard on the right side and wheeze and crackles on the left side. Cardiac examination revealed heart sounds on the right side without murmur. Apex beat was not visualised. Other systemic examination was within normal limits.

INVESTIGATIONS

The examination of blood revealed neutrophilic leucocytosis, with a haemoglobin value of 120 g/L (haematocrit 36.8%), total leucocyte count $13.2 \times 10^9/L$ (neutrophils $8.8 \times 10^9/L$, lymphocytes $3.8 \times 10^9/L$) and platelet count $175 \times 10^9/L$. Chest radiograph showed left hemithorax hyperlucency and hyperinflation with opaque right hemithorax

Figure 1 Photograph showing hypoplasia of thumb.

Figure 2 Chest x-ray showing left hemithorax hyperlucency with opaque right hemithorax and mediastinal shift.

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Figure 3 Colour Doppler study of heart showing coratrium.

and mediastinal shift to the right side. Bronchovascular markings were prominent on the left side (figure 2). ECG demonstrated a normal sinus rhythm pattern. Two-dimensional echocardiography with colour Doppler showed cor-triatrum with left to right shunt at an atrial level with mild pulmonary artery hypertension. QP:QS ratio was 2.2:1 (figure 3). A CT of chest showed no visualisation of right pulmonary artery and main pulmonary artery continuing as left pulmonary artery. The CT shows severely hypoplastic right lung with small-sized right bronchus and compensatory hyperinflation of left lung with transthoracic herniation towards the right side (figures 4 and 5). The thoracic cage showed spina bifida occulta of C7 vertebra and hemivertebra of D1 (figure 6). Renal and liver function tests were normal. Blood culture was sterile and ELISA for HIV was negative.

DIFFERENTIAL DIAGNOSIS

Lobar pneumonia is the most common diagnosis but the absence of air bronchogram in chest radiograph excludes this condition.

Figure 4 A CT scan showing no visualisation of right pulmonary artery with hypoplastic right lung and main pulmonary artery continuing as left pulmonary artery. The scan also shows spina bifida occulta of C7 vertebra and hemivertebra of D1.

Figure 5 A CT scan showing spina bifida occulta of C7 vertebra and hemivertebra of D1.

TREATMENT

The patient was treated conservatively with levosalbutamol nebulisation four times every hour, intravenous cefotaxime and cloxacillin for 10 days and improved symptomatically. At discharge, the patient was started on enalapril (0.1 mg/kg/day) for pulmonary hypertension and advised to follow-up in the outpatient department.

OUTCOME AND FOLLOW-UP

The patient is doing well in follow-up at 15 days and 3 months without any episode of lower respiratory tract infection. At 3 months, orthopaedic surgeon has advised for conservative management for vertebral anomalies as he had no neurological deficit. The patient had lower respiratory tract infection at

Figure 6 Three-dimensional image showing hemivertebra of D1.

1 year of follow-up, which was managed with oral bronchodilator and amoxicillin and clavulenic acid combination for 7 days.

DISCUSSION

UPAA was first reported by Fraentzel in 1868.² Thereafter review of 98 and 150 cases were published.^{3–4} Another review published in 2002, reported 108 cases of UPAA of the right or left pulmonary artery from 1978 to 2000.¹ UPAA results from the lack of embryological development of either the left or right sixth aortic arch and has various clinical presentations. In the absence of a single source of inflow, the pulmonary arterial circulation is derived from segmental vessels arising from the aorta, including a subphrenic contribution from the coeliac axis.⁵ The most common sign during childhood is contralateral pulmonary hypertension and in adults, haemoptysis.¹ Absent left pulmonary artery occurs more frequently in association with other cardiac malformations than right pulmonary artery agenesis.⁶ Vertebral anomaly in this patient may have resulted in hypoplastic thumb and thenar atrophy.

Long-standing mild dyspnoea and exercise intolerance are reported.⁷ Clinical examination may show asymmetry of the chest with ipsilateral deviation of the trachea, and decreased breath sounds on the involved side.

The chest radiographic appearance may be misinterpreted as Swyer-James syndrome, compensatory or obstructive emphysema and pulmonary embolism. Typical features include ipsilateral contracted lung with ipsilateral mediastinal and tracheal shift, elevation of ipsilateral hemidiaphragm and hyperinflation of contralateral lung.^{4–8}

Although angiography remains the gold standard for diagnosing UPAA⁸ it is rarely performed now unless embolisation is suggested by massive haemoptysis.⁹ Both CT and MRI can accurately depict the absent pulmonary artery.¹⁰ The presence of hilar arteries can be demonstrated by pulmonary venous wedge angiography.

When pulmonary hypertension is present in a patient with UPAA, their condition may be improved by revascularisation of the side with the absent artery. In the case of large collateral arteries leading to pulmonary hypertension or haemoptysis, embolisation techniques can be used. We have used enalapril in this patient. Enalapril by inhibiting tumour necrosis factor mediated activation of transcription factor NF- κ B and AP-1, which is required for α 1(I) collagen expression, decreases in lung inflammation, collagen deposition and muscle thickness in media of small pulmonary vessels, thereby causing decrease in pulmonary artery pressure.¹¹ It also attenuates fibro-proliferative

effects of chronic hypoxia and partially reverses the vascular neointimal formation and smooth muscle cell proliferation.

The early diagnosis of UPAA thus can prevent further deterioration and complications of bronchiectasis. There is an overall death rate of 7%.¹ Only a few patients with UPAA remain asymptomatic. UPAA may be suspected by the presence of recurrent respiratory infections, haemoptysis or pulmonary hypertension and of course by the typical chest radiograph findings.

Learning points

- ▶ Lower respiratory tract infection is common in children.
- ▶ Unilateral pulmonary artery agenesis (UPAA) is a rare cause of lower respiratory tract infection.
- ▶ UPAA with vertebral and cardiac anomaly is extremely rare.

Contributors UKS and RP drafted the manuscript and GNS and OPM managed the patient, carried out literature review and followed the patient.

Competing interests None.

Patient consent Obtained.

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