Unilateral right pulmonary artery agenesis and congenital cystic adenomatoid malformation of the right lung with Ortner’s syndrome

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ABSTRACT

We report a 2.5-year-old girl who presented with hoarseness of voice since 3 months of age and failure to thrive. Chest X-ray showed cardiomegaly with a deviation of the trachea and mediastinum to the right side. Two-dimensional echocardiography showed decreased flow across the right pulmonary artery, a small atrial septal defect (ASD) with a right-to-left shunt, and a dilated right atrium and right ventricle with severe tricuspid regurgitation suggestive of severe pulmonary hypertension. A silent large patent ductus arteriosus was also seen. Multiple detector computerized tomography aortogram confirmed the findings of absent right pulmonary artery and hypoplastic right lung with small cystic lesions suggestive of congenital cystic adenomatoid malformation in the right lower lobe. Hoarseness of voice was due to the left vocal cord palsy probably secondary to severe pulmonary hypertension (Ortner’s syndrome).

KEY WORDS: Congenital cystic adenomatoid malformation of lung, Ortner's syndrome, patent ductus arteriosus, pulmonary hypertension, unilateral absence of pulmonary artery

INTRODUCTION

Unilateral absence of pulmonary artery (UAPA) is a rare congenital abnormality, with an estimated prevalence of 1 in 200,000.[1] Some patients with UAPA are totally asymptomatic while others may have severe pulmonary hypertension, hemoptysis, congestive heart failure, and cyanosis.[1] We report a 2.5-year-old girl with congenital absence of the right pulmonary artery with associated congenital cystic adenomatoid malformation (CCAM) of the right lower lobe, patent ductus arteriosus (PDA), and atrial septal defect (ASD), who presented with Ortner’s syndrome due to severe pulmonary hypertension.

CASE REPORT

A 2.5-year-old girl presented with hoarseness of voice noticed since 3 months of age, breathlessness for the past 15 days and failure to thrive. She had been admitted elsewhere for lower respiratory tract infection at 3 months of age. An otorhinolaryngology reference for hoarseness of voice had revealed left vocal cord palsy. The child was noticed to have a weak cry and an exaggerated suck-rest-suck cycle and sweating over the forehead while feeding. Examination revealed a cyanosed child with Grade 2 clubbing. Respiratory rate was 60/min with respiratory

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distress. Pulse rate was 160/min. All peripheral pulses were well felt. Blood pressure in the right upper limb was 94/60 mmHg. Pulse oximetry in room air showed an oxygen saturation varying between 60% and 70% in all four limbs. Weight and height were 7 kg and 77 cms, respectively both below the third percentile for age and sex. There were no dysmorphic features.

Examination of the cardiovascular system revealed a precordial bulge with visible epigastric pulsations. An ejection systolic murmur Grade 3 was heard in the right parasternal region in the fourth and fifth intercostal spaces. Examination of the respiratory system revealed diminished breath sounds over the right hemithorax. The liver was palpable 2 cm below the right costal margin in the mid-clavicular line.

The complete blood count was essentially normal. Chest X-ray [Figure 1] revealed cardiomegaly with a cardio-thoracic ratio of 0.6 and deviation of the trachea and mediastinum to the right. There was mild hyperinflation of the left lung. Arterial blood gases showed hypoxemia with oxygen saturation of 68%. Electrocardiogram showed sinus tachycardia with P pulmonale. Two-dimensional (2D) echocardiography showed decreased flow in the right pulmonary artery, a small ASD with a right-to-left shunt, and a dilated right atrium and right ventricle with severe tricuspid regurgitation, suggestive of severe pulmonary hypertension. A silent large PDA was also seen. Multiple detector computerized tomography aortogram [Figure 2] confirmed the findings of ASD with pulmonary hypertension, absent right pulmonary artery and hypoplastic right lung with small cystic lesions suggestive of CCAM in the right lower lobe. Blood culture was sterile.

At admission, the patient was started on oxygen, furosemide, enalapril and intravenous cefotaxime, on a provisional diagnosis of congenital cyanotic heart disease with increased pulmonary blood flow, lower respiratory infection, and congestive cardiac failure. Despite these measures, there was persistent hypoxemia and worsening of cardiac failure in the form of tachycardia and bilateral basal crepitations. The child was intubated and ventilated and started on pressors, but sustained a cardiac arrest on the 4th hospital day from which she could not be resuscitated.

**DISCUSSION**

Complete arrest of pulmonary artery supply results in the arrest of early bronchial development leading to agenesis of the affected lobe or segment. Pulmonary agenesis is usually unilateral, right sided absence of pulmonary artery being more common. More than 50% of children with pulmonary agenesis have associated congenital anomalies that involve the cardiovascular (PDA and patent foramen ovale), gastrointestinal, skeletal, and genitourinary systems. Our patient had ASD and PDA. While some patients with UAPA are totally asymptomatic, others may have severe pulmonary hypertension, hemoptysis, congestive heart failure, and cyanosis. Chest X-ray may show an absent hilar shadow, a shrunken affected lung, and shift of the mediastinum to the affected side. Diagnosis may be confirmed on 2D echocardiography.

Our patient had additionally CCAM of the right lower lobe. An early interruption in the development of the pulmonary artery could result in the continued development of the primitive capillary supply to a region of the lung with resultant abnormal development of the supplied region. The magnitude of the insult would determine the exact development of the affected lung tissue and final blood supply, resulting in pulmonary sequestration, CCAM, or a combination of the two lesions.

The incidence of pulmonary hypertension in patients with isolated UAPA varies between 18% and 44%. In patients

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**Figure 1:** Chest X-ray posterioranterior view showing cardiomegaly and shift of trachea and mediastinum to the right

**Figure 2:** Multiple detector computerized tomography aortogram showing absent right pulmonary artery, and cystic lesions in the right lower lobe suggestive of congenital cystic adenomatoid malformation (arrow) of lung
with UAPA with PDA as in our patient, the incidence of pulmonary hypertension was observed to be as high as 86%, those with pulmonary hypertension dying at an early age.\[^{[5]}\]

Our patient presented initially as Ortner’s syndrome at the age of 3 months. Ortner syndrome or cardiovocal syndrome refers to hoarseness of voice due to recurrent laryngeal nerve paralysis secondary to cardiovascular disease. This syndrome was first described by Ortner in 1897 in two patients who had mitral stenosis and left recurrent laryngeal nerve paralysis.\[^{[6]}\] The syndrome has since been described in adults with various cardiovascular disorders, but reports in children are less common.\[^{[7,8]}\] The pulmonary artery, enlarged due to pulmonary hypertension, has been implicated as the main mechanism of nerve injury. Fetterolf and Norris studied the anatomic relations of the left recurrent laryngeal nerve in cadavers and concluded that the nerve must be squeezed between the enlarged left pulmonary artery and the aorta or ligamentum arteriosum.\[^{[9]}\]

Unilateral absence of pulmonary artery should be considered in the differential diagnosis of children who present with cyanosis, pulmonary hypertension, or recurrent lower respiratory tract infections.

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**Conflicts of interest**

There are no conflicts of interest.

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