Case Report

Pulmonary Agenesis

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Abstract

Unilateral opaque lung with ipsilateral mediastinal shift is an uncommon cause of respiratory distress in newborn which can be found on simple radiograph of the chest. Pulmonary agenesis is a rare cause of unilateral opaque lung in the newborn. Nearly 50% cases of pulmonary agenesis are associated with other congenital defects including cardiovascular, skeletal, gastrointestinal or genitourinary systems.[1] We report an infant with agenesis of the right lung associated with other congenital anomalies. **[Indian J Chest Dis Allied Sci 2015;57:251-253]**

Key words: Pulmonary agenesis, Hypoplasia, Respiratory distress, Newborn.

Introduction

Although the exact prevalence of pulmonary agenesis is not known, it has been reported between 34 to 97 cases per 10 lakh live births.[2] Pulmonary agenesis, aplasia and hypoplasia are few congenital abnormalities of the lung that are included in the differential diagnosis of unilateral opaque lung with ipsilateral mediastinal shift in the newborn and children. Adults are rarely affected. Depending on the stage of development of the primitive lung bud, pulmonary agenesis can be classified into three categories[3]: (i) agenesis — complete absence of pulmonary tissue and bronchial tree with no vascular supply to the affected lung; (ii) aplasia — rudimentary bronchus and pulmonary artery are present, but there is complete absence of pulmonary parenchyma; and (iii) hypoplasia — presence of variable amounts of bronchial tree, pulmonary parenchyma and supporting vasculature. Clinically it presents as shortness of breath, wheezing, tachypnoea, repeated respiratory tract infections and respiratory distress. We present the case of a newborn girl with unilateral pulmonary aplasia of the right lung found during the work-up for respiratory distress.

Case Report

A full-term (40 weeks) woman with polyhydramnios gave birth to a female child. Baby was delivered by normal vaginal delivery but the condition of the baby at birth was critical. There was no spontaneous gasping or crying; she was not responding to painful stimuli; and her heart rate was <100 per minute. There was no spontaneous breathing effort. She was intubated and put on mechanical ventilation. A chest radiograph showed right-sided opaque hemithorax with ipsilateral mediastinal shift and herniation of left lung (Figure 1).

Echocardiogram and colour Doppler studies showed situs solitus, heart was in the right hemithorax (dextrocardia). There was a large ostium secundum atrial septal defect (6 mm) with left-to-right shunt. Also,

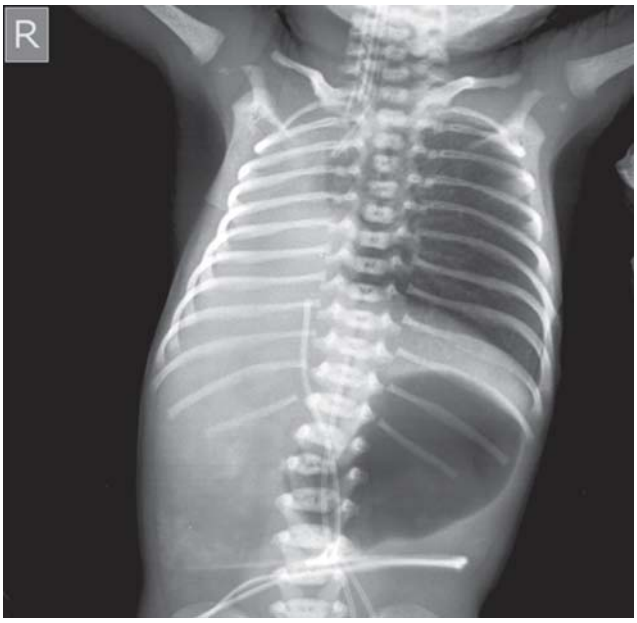

Figure 1. Chest radiograph showing completely opaque right hemithorax with ipsilateral mediastinal shift and herniation of the left lung.

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there was a large sub-aortic ventricular septal defect. Aorta and pulmonary artery lay side by side with both the great vessels exiting from the right ventricle. There was no coarctation of aorta or patent ductus arteriosus. Dilation of the right atrium and ventricle was seen with small left atrium and ventricle.

Contrast-enhanced computed tomographic scan of the chest showed completely opaque right hemithorax with no evidence of right pulmonary artery or bronchial tree or pulmonary parenchyma, with complete ipsilateral shift of mediastinal structures to the right. Compensatory hyperinflation of the left lung was seen with herniation into the right hemithorax. Left pulmonary artery, left main-stem bronchus and lung parenchyma were normally visualised (Figure 2). Trachea was seen with endotracheal tube and oesophagus was seen with feeding tube *in situ*. Walls between mid oesophagus and adjacent trachea/left main bronchus were not well visualised giving the impression of a tracheo-esophageal fistula (Figure 3).

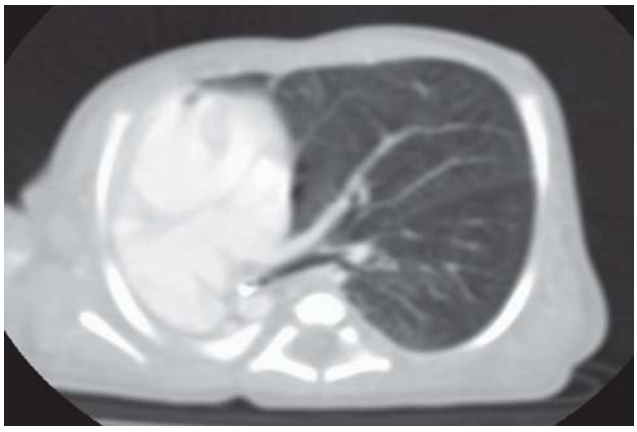

Figure 2. Computed tomography of the chest showing absence of the right lung; left pulmonary artery, bronchial tree and well-formed parenchyma.

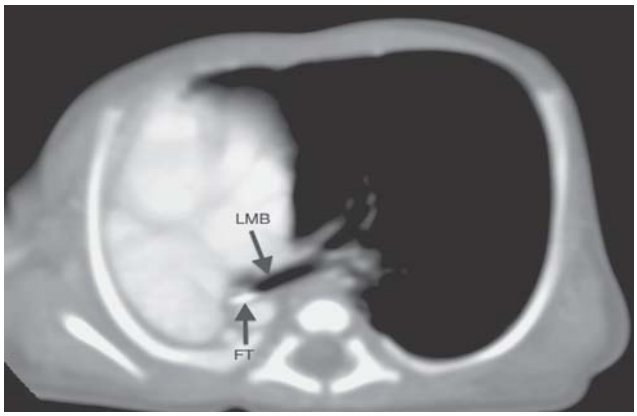

Figure 3. Computed tomography of the chest showing probable tracheo-esophageal fistula. The area between mid oesophagus and adjacent left main bronchus are not visualised well.

LMB=Left main bronchus; FT=Feeding tube.

Discussion

Pulmonary agenesis, aplasia and hypoplasia have been reported at different ages in newborns, infants, children and adults.[4] Bilateral pulmonary agenesis is a rare malformation that may be associated with anencephaly and is incompatible with life.[5] Unilateral agenesis, aplasia and hypoplasia are comparatively more common and may have few symptoms and non-specific findings, among which only one-third are diagnosed during life.[6]

Embryologically, these malformations correspond to a failure of the development of the respiratory system from the foregut. Arrest at the stage of the primitive lung bud produces bilateral pulmonary agenesis. Lobar agenesis results when developmental arrest occurs on one side in an older embryo. Pulmonary hypoplasia may occur during the last trimester of pregnancy with failure of final alveolar differentiation.[5] Pulmonary agenesis and aplasia are rare abnormalities thought to have an incidence between 0.0034% and 0.0097%.[2] Genetic, teratogenic and mechanical factors may have a bearing on aetiology.[6] These are generally sporadic, with only a few reports of these conditions occurring in siblings in an autosomal recessive pattern. These occur with equal frequency in both the sexes and involve both lungs equally.[4] Nearly 50% cases of pulmonary agenesis have associated congenital defects,[7] which usually involve cardiovascular, skeletal, gastrointestinal and genitourinary system. Congenital absence of breast is rare. The VACTERL association (also VATER syndrome) refers to the non-random co-occurrence of birth defects, namely vertebral anomalies, anal atresia, cardiac defects, tracheo-esophageal fistula and/or oesophageal atresia, renal anomalies and limb defects. In the present case there was a combination of pulmonary agenesis, cardiac defects and tracheo-esophageal fistula.

The exact aetiology of this condition is unknown though genetic factors, viral agents and dietary deficiency of vitamin-A during pregnancy have been implicated. Left-sided agenesis is more common and the subjects have a longer life expectancy than those with the right-sided agenesis. This is probably due to excessive mediastinal shift and malrotation of carina in the right-sided agenesis which hinders proper drainage of the functioning lung and increases the chance of respiratory infections.[8]

Previously, diagnosis of pulmonary agenesis used to be made by chest radiography, bronchography, bronchoscopy and angiography. After the invention of computed tomographic scan, these invasive procedures which carry a significant risk have gone out of vogue.[9]

Prognosis depends on two factors: (i) severity of associated congenital anomalies, and (ii) involvement of the normal lung in any disease process.[10] Patients with

right lung agenesis have a higher mortality because of compression of the tracheo-bronchial tree by the shifting of normally mid thoracic structures into the right chest.[11] If patient survives the first five years without major infection, an almost normal life span can be expected.[7]

References

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