Indian Journal of Medical Case Reports ISSN: 2319–3832(Online)
An Open Access, Online International Journal Available at http://www.cibtech.org/jcr.htm
2014 Vol.3 (2) April-June, pp.48-51/Kapoor et al.
Case Report

LEFT SIDED PULMONARY AGENESIS WITH CONGENITAL HEART ANOMALIES AND MILD PULMONARY HYPERTENSION: A RARE CASE REPORT

***Rohit Kapoor, Rakesh Yadav, Premila Paul and Soumya Sachdeva**
Department of Pediatrics, Vardhman Mahavir Medical College and Safdarjang Hospital
**Author for Correspondence*

ABSTRACT

Background: Pulmonary agenesis is the complete absence of bronchus, parenchyma and vessels. Unilateral lung agenesis is more common (Aggarwal *et al.,* 2002). Accumulation of bronchial secretions and products of inflammation are chief determinants that determine the degree of respiratory distress (Krivchenya *et al.,* 2000). Anomalies frequently associated with pulmonary agenesis may include any organ system viz, cardiovascular, skeletal, gastrointestinal and genitourinary systems (Krivchenya, 2000; Nazir *et al.,* 2006; Kaya and Dilmen, 1989; Lucaya and Strife 2002). These are more commonly found in association with right pulmonary agenesis. Vascular aberrations and genitourinary system malformations have also been noted with the presence of right pulmonary agenesis. Left pulmonary agenesis is often an isolated anomaly; associated with usually no congenital defects (Maltz and Nadas, 1968). In our case; despite left pulmonary agenesis, there was evidence of atrial septal, ventricular septal defect, mild pulmonary hypertension with antenatally diagnosed bilateral renal hypoplasia.

Keywords: *Pulmonary Agenesis, Congenital Anomaly, Recurrent Pneumonia*

CASES

A 7-month old infant born to non- consanguineous parents presented with history of fever, cough, and respiratory distress, with decreased oral acceptance since 2 weeks. History revealed multiple episodes of chest infections since 3 months of age and episodes were treated symptomatically for the exacerbations as bronchopneumonia; with bronchodilators and antibiotics. Child had multiple episodes of bronchopneumonia managed on the lines of recurrent pneumonia. Mother never complained of cyanosis, however child was never asymptomatic in between the intervals and always suffered from distress. Antenatal checkups were normal and no history of oligohydramnios was elicited.The full-term child was born through normal vaginal delivery, cried immediately after birth and developed respiratory distress; treated conservatively and discharged uneventfully. The height, weight and length of the child was normal compared to boston standards. Examination revealed drooping of shoulder with crowding of ribs and decreased chest expansion on the left side; and scattered rales and bronchial breath sounds and increased vocal resonance on the right side. Apex beat was displaced to the 5th intercostal space and heart sounds revealed a pansystolic murmur over the mitral area. Investigations demonstrated a hemoglobin of 10.7gm/dl, total leukocyte count of 14,300/mm^3 with predominance of polymorphs 78%.Rest of the differential was constituted by lymphocytes 13%, eosinophils 4% and monocytes 5%. The findings noted on chest x ray included opacity in the left lung fields and scattered infiltrates on the right side with compensatory hyperinflation and a increased cardiothoracic ratio of 0.63 (figure1).

Owing to severe respiratory distress; the child was admitted and rescue medications, oxygen, intravenous fluids and antibiotics were begun. On bronchoscopy; left lung tissue was visualized upto segmental bronchi indicating left lung agenesis. Left pulmonary artery and vein were not visualized, which was later confirmed by pulmonary angiography. Echocardiography showed atrial and ventricular septal defect with cardiomegaly with a increased CT ratio; with features of mild pulmonary hypertension. The contrast study of chest revealed lung agenesis with a compensatory mediastinal shift to the left, hyperinflation involving right lung and prominent pulmonary vasculature (figure2).

© Copyright 2014 | Centre for Info Bio Technology (CIBTech)

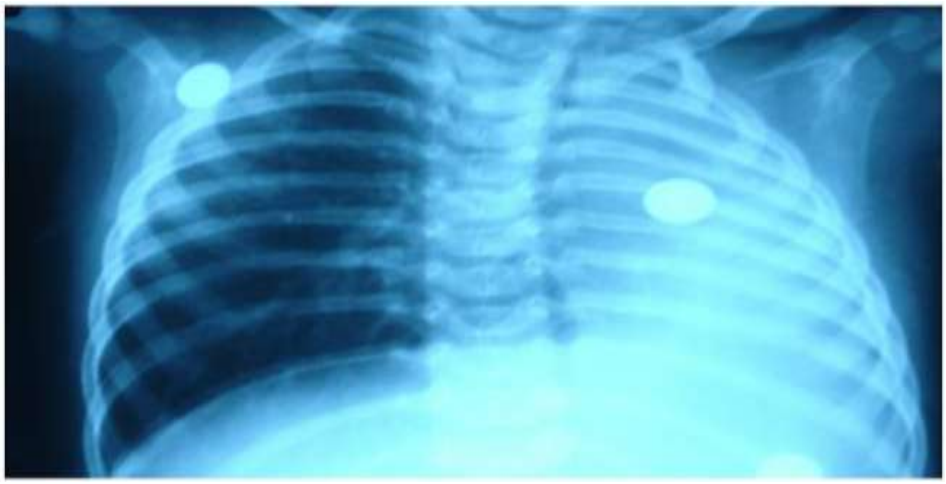

Figure 1: Chest X ray AP view showing left sided opacity and left sided compensatory hyperinflation with few scattered infiltrates

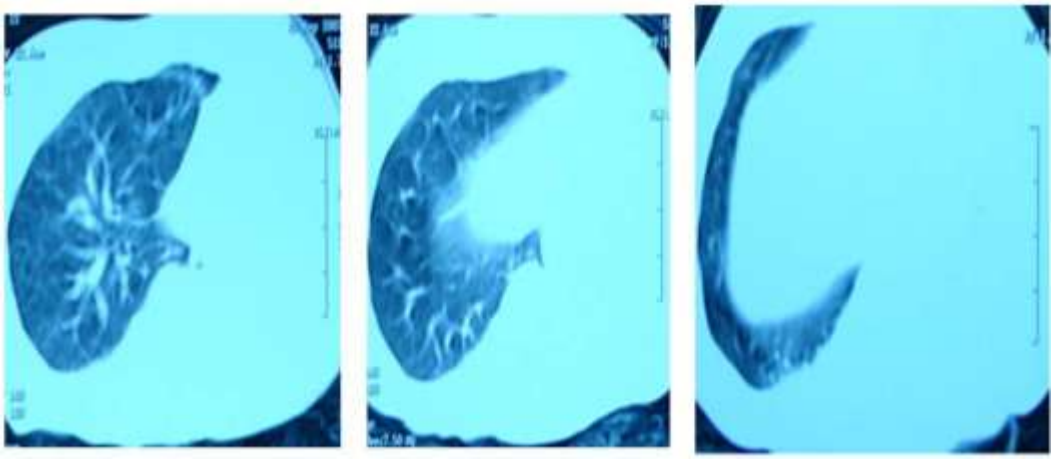

Figure 2: Sequential horizontal section showing left lungs agenesis with mediastinal shift to left side with overinflation of right lung. Infiltrates can be appreciated in right lung fields

The sonogram of the abdomen showed hypoplasia of the kidneys however the renal function tests were high normal with BUN 40mg% and creatinine of 1.2mg%. Also the child had a normal urine output. This patient though having left lung agenesis; had presence of concomitant cardiovascular anomalies and features of mild pulmonary hypertension, and bilateral renal hypoplasia; which is an unusual association. This patient inspite of having left lung agenesis; had cardiovascular anomalies in form of septal defects and antenatally diagnosed defect with features of mild pulmonary hypertension and bilateral renal hypoplasia.

DISCUSSION

The prevalence of lung agenesis is reported to be 34 per 1,000,000 live births (Mardini and Nyhan, 1985). It's a rare condition and can be can involve single or both lungs. Systemic malformations complicating unilateral lung agenesis are the usual cause of death after birth; which may occur in early childhood or late adulthood. Left pulmonary agenesis is often manifested without other congenital defects; frequently associated anomalies may include any organ system viz, cardiovascular, skeletal, gastrointestinal, genitourinary systems (Krivchenya, 2000; Nazir *et al.*, 2006; Kaya and Dilmen, 1989; Lucaya and Strife 2002; Maltz and Nadas, 1968).

© Copyright 2014 | Centre for Info Bio Technology (CIBTech)

Case Report

Pulmonary agenesis usually occurs between the 4th and 5th week of gestation during the embryonic phase, before the pseudo glandular period, during which primitive lung forms a diverticulum protruding from the foregut. The chromosomal basis of the aberration is suggested to be related to duplication of the distal part of the upper arm of chromosome 2 (Say *et al.,* 1980). Authors of various studies have reported cases of pulmonary aplasia with dextro cardia (Nandan *et al.,* 2012); case of deviated trachea and aplasia of right lung (Dohlemann *et al.,* 1990). Unilateral tracheal stenosis along with, unilateral pulmonary agenesis and patent ductus arteriosus have been mentioned in the literature (Nelson *et al.,* 1967) and can rarely occur as a part of VACTERYL sequence (Knowles *et al.,* 1988). A rare case of bilateral pulmonary agenesis, oesophageal atresia and first arch syndrome has been reported as well (Buse and Morris, 1973). Hemifacial microsmia has also been reported in patients with this anomaly (Panigrahi *et al.,* 2010). Lung aplasia has also been reported along with hereditary renal dysplasia and mayer-rokitansky-kusterhauser syndrome (Acién *et al.,* 2010).

Children suffering from this disorder present with recurrent respiratory infections, cough, retractions, wheezing, tachypnoea from early age onwards (Mardini and Nyhan, 1985).

Bilateral lung agenesis is a grave, life threatening condition with a 5 year survival rate of about 50%. patients. Patients with left lung agenesis having better prognosis than the right one (Dohlemann *et al.,* 1990). The absence of critical anomalies in patients with left lung agenesis may even be the cause of increased survival in this set of patients. According to Say *et al.,* (1980) Radiology plays a crucial rule; modalities like chest x ray, bronchography, bronchoscopy, and contrast angiography are used to gain leverage and to visualize the characteristic findings showing opaque hemithothorax, and clearly delineating lung parenchyma, pulmonary and bronchial tree (Dohlemann *et al.,* 1990).

Diagnosis of this condition is made on the basis of characteristic contrast findings which show opaque hemithorax; which clearly delineates lung parenchyma, pulmonary and bronchial tree (Dohlemann *et al.,* 1990).

In our case, the child presented immediately after birth with respiratory distress that resolved on its own, however the multiple episodes of chest infections were misdiagnosed as recurrent pneumonia. The unique characteristic of our case was that the child had atrial and ventricular septal defects with features of mild pulmonary hypertension and bilateral renal hypoplasia; in association with left pulmonary agenesis. These anomalies are however found more commonly in case of right lung agenesis.

Conclusion

Thus, in cases of repeated chest infections associated with left lung agenesis, looking for other rare congenital anomalies is imperative and must be kept in mind. Asymptomatic cases do not require any treatment, if there are no life threatening or additional anomalies.

REFERENCES

Aggarwal AN, Gupta D and Jindal SK (2002). Unilateral pulmonary agenesis with ipsilateral musculoskeletal anomalies. *Journal of the Association of Physicians of India* **50** 1324–1326.

DeBuse PJ and Morris G (1973). Bilateral pulmonary agenesis, oesophageal atresia, and the first arch syndrome. *Thorax* **28**(4) 526–528.

Devki Nandan, Girish Chandra Bhatt, Vivek Dewan and Imkongkumzuk Pongener (2012). Complete Right Lung Agenesis with Dextrocardia: An Unusual Cause of Respiratory Distress. *Journal of Clinical Imaging Science* **2** 81. doi: 10.4103/2156-7514.105140

Dohlemann C, Mantel K, Schneider K, Guntner M, Kreuzer E and Hecker WC (1990). Deviated trachea in hypoplasia and aplasia of the right lung: Airway obstruction and its release by aortopexy. *Journal of Pediatric Surgery* **25** 290–3.

Inusha Panigrahi, Rashmi Ranjan Das, Ram Kumar Marwaha (2010). Hemifacialmicrosomia with pulmonary hypoplasia. *BMJ Case Reports* **2010**(May 17). doi: 10.1136/bcr.04.2009.1759

Kaya IS and Dilmen U (1989). Agenesis of the lung. *European Respiratory Journal* **2** 690–2.

Knowles S, Thomas RM, Lindenbaum R, Keeling JW and Winter RM (1988). Pulmonary agenesis as part of the VACTERL sequence. *Archives of Disease in Childhood* **63**(7 Spec No) 723–726.

© Copyright 2014 | Centre for Info Bio Technology (CIBTech)

Krivchenya DU, Dubrovin AG, Krivchenya TD, Khursin VN and Lysak CV (2000). Aplasia of the right lung in a 4-year-old child: Surgical stabilization of the mediastinum by diaphragm translocation leading to complete recovery from respiratory distress syndrome. *Journal of Pediatric Surgery* **35** 1499–502.

Lucaya J and Strife J (2002). *Pediatric Chest Imaging: Chest Imaging in Infants and Children*. Edited by Lucaya J and Strife J (Berlin, Germany: Springer-Verlag) 93–112.

Maltz DL and Nadas AS (1968). Agenesis of the lung: presentation of eight new cases and review of the literature. *Pediatrics* **42** 175–188.

Mardini MK and Nyhan WL (1985). Agenesis of the lung: Report of four patients with unusual anomalies. *Chest* **87** 522–7.

Meier M (2000). Agenesis of the left lung in combination with a persistent left superior vena cava and a cortriatriatum sinister in a 77-year-old man. *Pneumologie* **54** 249–51.

Nazir Z, Qazi SH, Ahmed N, Atiq M and Billoo AG (2006). Pulmonary agenesis-vascular airway compression and gastroesophageal reflux influence outcome. *Journal of Pediatric Surgery* **41** 1165–9.

Nelson CS, McMillan IKR and Bharucha PK (1967). Tracheal stenosis, pulmonary agenesis, and patent ductusarteriosus. *Thorax* **22**(1) 7–12.

Pedro Acién, Francisco Galán, Irene Manchón, Eva Ruiz, Maribel Acién, Luis A Alcaraz (2010). Hereditary renal adysplasia, pulmonary hypoplasia and Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome: a case report. *Orphanet Journal of Rare Diseases* **5** 6. doi: 10.1186/1750-1172-5-6

Say B, Carpenter NJ, Giacoia G and Jegathesan S (1980). Agenesis of the lung associated with a chromosome abnormality (46, XX, 2p+). *Journal of Medical Genetics* **17** 477–8.

© Copyright 2014 | Centre for Info Bio Technology (CIBTech)