CASE REPORT

Agenesis of the Lung-a Rare Congenital Anomaly of the Lung

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Abstract- Pulmonary agenesis is a very rare condition and many of them are associated with a variety of cardiac and non-cardiac malformations. We report an eight-month old girl with chronic lung infection due to right sided pulmonary agenesis without any associated major cardiac or non-cardiac abnormalities. The case brings in forth the importance of investigating any infant presenting with features of chronic lung infection for any congenital abnormality of the lung including aplasia of the lung. This case also emphasizes that mildness of the attack does not exclude right sided aplasia of the lung.

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Keywords: Consanguinity; Pulmonary agenesis; Rudimentary bronchus

Introduction

Pulmonary agenesis is an extremely rare condition with a reported prevalence of 34 per 10 lakh live births (1). Half of the cases have associated congenital malformation of the cardio-vascular, skeletal, gastro-intestinal or genitor-urinary system (2). Consanguinity has been reported and autosomal recessive may be the mode of inheritance (3). The first case from India was reported by Muhamed (4) in 1923 in a medico legal autopsy which showed left sided pulmonary agenesis. The most exhaustive reviews are those of Oyamada *et al.* (5).

Case Report

An eight month old Hindu female baby was admitted to the pediatric ward for the evaluation of respiratory distress, cough and wheeze for last 5 months. She was delivered by caesarian section and postnatal period was uneventful. She had an acute exacerbation of respiratory distress for last 12 days before admission. There was no history of consanguinity, oligohydramnios or prolonged premature rupture of the membrane. She weighed 6 kg, her pulse rate was 120/minute and respiratory rate was 60/minute. Chest examination showed decreased breath sound on the right side. Cardiovascular system examination revealed displacement of the apex to the right side. There was no bony or vertebral defect. Chest x-ray showed opacity at the right side of the chest with herniation of left lung to the right side (Figure 1).

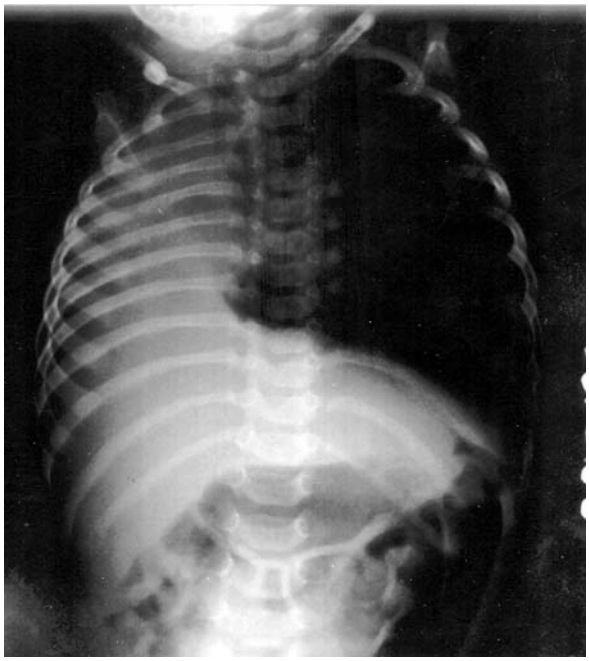

Figure 1. Chest x-ray of the patient showing opacity at the right side of the chest with herniation of left lung to the right. Mediastinum with heart is displaced to the right.

Mediastinum with the heart was displaced to the right. CT scan of the chest showed absence of right lung (Figure 2). Brochoscopy revealed obliteration of the right bronchus just after the carina. Left lung appeared to be normal. Rigid bronchoscope (3 mm) could not be negotiated into the right bronchus. Electrocardiogram and ultrasonography of the abdomen were normal.

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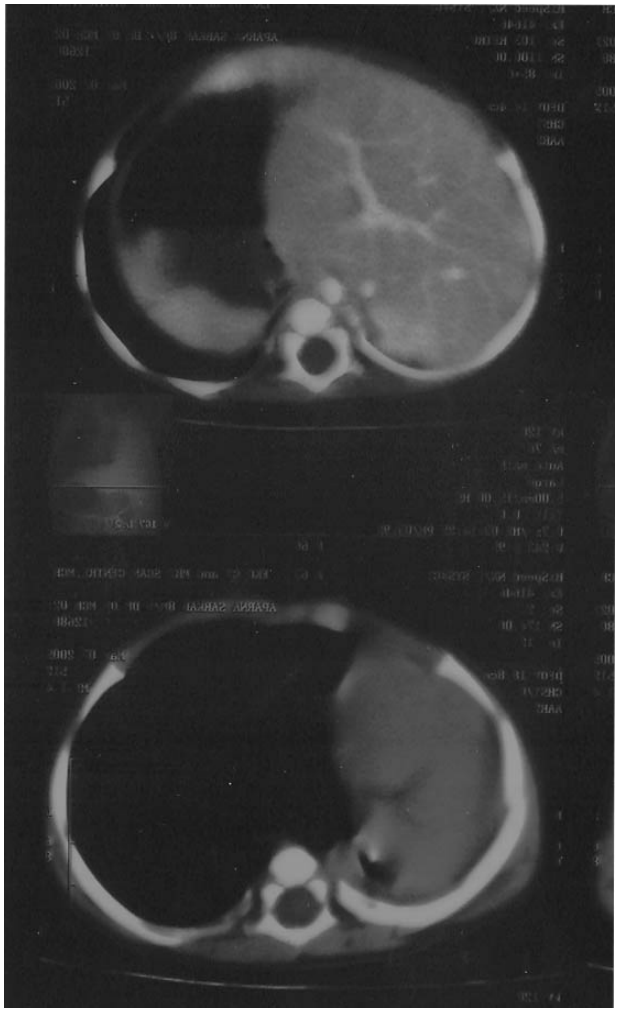

Figure 1. CT scan of the chest showing absence of right lung.

Echocardiogram revealed mild atrial septal defect. Right pulmonary artery could not be located. The twin was healthy and her chest x-ray was normal.

The case was diagnosed to have agenesis of the right lung (grade II of Schneider's classification). No other congenital anomalies were identified except mild atrial septal defect.

Discussion

Agenesis of lung is a rare congenital disorder (1). Left sided pulmonary agenesis is more common and these subjects have a longer life expectancy than those with right sided pulmonary agenesis (5). This is probably due to excessive mediastinal shift and malrotation of carina in right sided agenesis which hinders proper drainage of the functioning lung and increased chance of respiratory infections (6). Our patient was a female child with right sided pulmonary agenesis without any history of consanguinity or any associated malformation of the vertebra, ribs or thumb. The differential diagnoses of the condition included collapse, foreign body aspiration, bronchial mass lesion, thickened pleura and pneumonectomy. Malformations of the thumb, vertebra and costal defect have been reported by Booth *et al.* (7). Among the associated lesion, renal agenesis and *in situ* neuroblastoma of the adrenal gland are also reported (8). A wide variety of congenital cardiac abnormality associated with pulmonary agenesis have been reported including tricuspid regurgitation, tetralogy of Fallot, total anomalous pulmonary venous drainage, patent ductus areriosus and atrial septal defect (9). In our case only mild atrial septal defect was detected by echocardiography and no genito-urinary defect was found by ultrasonography of the abdomen.

Schneider (10) classified agenesis into three groups which has been subsequently modified by Boyden (11). Depending upon the stage of development of the primitive lung bud, pulmonary agenesis is classified into three categories:

Type 1 (Agenesis): Complete absence of lung and bronchus and no vascular supply to the affected side,

Type 2 (Aplasia): Rudimentary bronchus with complete absence of pulmonary parenchyma.

Type 3 (Hypoplasia): Presence of variable amounts of bronchial tree, pulmonary parenchyma and supporting vasculature.

Our patient would belong to type 2 pulmonary agenesis.

The onset of symptoms in pulmonary agenesis is remarkably variable. In many cases, presence of this anomaly usually comes to light during infancy because of recurrent chest infections, cardiopulmonary insufficiency or due to associated congenital anomalies. However, patients with one lung have been reported to survive well into adulthood without much complaint (6). Agenesis or aplasia of the lung can be managed medically and surgery is seldom required (6). .The oldest patient cited by Oyamada et al (5) was 72 years old.

Right sided agenesis has been thought to have a graver prognosis than that of left (5) with death occurring earlier and with increasing frequency. Many authors have suggested cardio-angiography for proper delineation of the pulmonary artery (6), but due to unavailability of the facility, we were contained in our case with echocardiography only which failed to demonstrate the right pulmonary artery .

By 1977, over 200 cases of under-development of lung have been reported (6). Our case is definitely an

important addition to this pool of anomaly. Moreover, though it is a right sided agenesis, the case has few problems except one episode of exacerbation. No consanguinity was documented and twin sister was healthy without any anomaly of the lungs. The case improved following medical therapy with antibiotics and oxygen. She is being followed up regularly at our out-patient clinic. In conclusion, agenesis of lung can be the cause of chronic lung infection during infancy and right sided agenesis of lung may be mild type too during the initial period of life.

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