UAPM
Journal of Respiratory Diseases
and Allied Sciences

Case Report

Lobar Agenesis with Cross Ectopic Lung in a Middle-Aged Male Masquerading Sequels of Pulmonary Tuberculosis: Extremely Rare and Challenging Case for Clinician

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Abstract

Pulmonary agenesis is a rare congenital anomaly. It usually presents in childhood with high mortality due to the involvement of other systems of the body. Here, we report a case of a 53-year-old male presented with left upper lobe pulmonary type 2 lobar agenesis complicated with recurrent pulmonary infection masquerading sequels of pulmonary tuberculosis.

Keywords: Agenesis, Aplasia, Congenital anomaly, Lung.

Introduction

Lung agenesis is a rare congenital phenomenon. The first case of lung atresia and agenesis was reported in 1673 through a medical autopsy in a female patient by De Pozze[1] and the first clinical diagnosis was made by Meyer M. in 1885.[2] Though the first case of unilateral left lung agenesis from India was reported by Muhamed K.S.N. in 1923 in a medico-legal autopsy.[3] According to literature, over 200 cases of lung dysplasia have been promulgated, though only 66 cases of unilateral lung agenesis in adulthood have been reported to date. The oldest patient cited in literature was 72 years old reported by Oyamada *et al.* in 1953.[4] The incidence of congenital pulmonary dysplasia is between 30 to 42 per 100,000 individuals moreover, lobar agenesis is rarest in reported cases, incidence of the disease is 0.0034 to 0.0097% with no gender predisposition.[5] Left lung agenesis is more common than right lung.[6] The average life expectancy of right lung agenesis was reported as 6 years, though it is 16 years for left lung agenesis,[7] and nearly 50% of patients have multi-system involvement.[8,9] Bilateral lung agenesis is incompatible with life and around 50% of affected individuals usually die within the first 5 years.[10,11]

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Case Report

A 53 year old male presented to the respiratory department with chief complaints of productive cough and mucopurulent sputum, breathlessness on exertion associated with chest pain for the last month, also complaining of low-grade fever with body aches off and on for last year. He had taken anti-tubercular treatment thrice in the past, though evidence of bacteriological confirmation was lacking in the provided record. On general examination, he has clubbing of grade 2. Cyanosis was absent, No evidence of icterus, pallor or pedal edema. BP 146/84 mm Hg, respiratory rate 22/minute, pulse rate 84/Minute, SPo2 91% at room air, temperature 101 Degrees F. Chest examination revealed tracheal shifting towards the left side with crowding of ribs over left hemi thorax. The apex beat shifted higher and more towards the anterior axillary line on left side. Air entry was diminished over the left upper zone compared to the contralateral site

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How to cite this article: Agrawal A, Singh K, Kharb S, Goel S. Lobar Agenesis with Cross Ectopic Lung in a Middle-Aged Male Masquerading Sequels of Pulmonary Tuberculosis: Extremely Rare and Challenging Case for Clinician. UAPM J. Respiratory Diseases Allied Sci. 2024;1(1):20-23.
Received: 15-01-2024, **Accepted:** 19-05-2024, **Published:** 25-08-2024

with the presence of bilateral wheezing sounds. Examination of other systems was unremarkable. Haemoglobin 14 gm%, Red blood count 4.5 × 10^6 mm^3, mean corpuscular volume 94 fl, mean corpuscular haemoglobin 31.2 pg, mean corpuscular haemoglobin concentration 33.2 gm/dl, Platelets count 2 lac/mm^3 total leucocyte count 10000/mm^3, differential leucocyte count polymorph 80%, lymphocytes 14%, monocytes 3%, eosinophils 3%. Aspartate transaminase/ Alanine aminotransferase 91/178 IU/dl serum urea 20 mg/dl, S. creatinine 0.9 mg/dl, C-reactive protein 24 IU/dl, Sputum smear for acid fast bacilli by Z-N staining and CBNAAT for mycobacteria was negative. Arterial blood gas analysis was within normal range except for mild hypoxemia (pH 7.4, Pco2 34 mm Hg, Po2 64.3 mmHg, HCO3 24 mmol/l). IL6 7.69 pg/ml, D-dimer 409 ng/mL, LDH 886 U/L, CPKMB 16 IU/L, PTINR 1.19. His skiagram of the chest in PA view showed left opaque hemithorax with shifting of mediastinum towards the left side, raised left hemi diaphragm (Figure 1). Contrast-enhanced computer tomography thorax in lung and mediastinal window revealed herniation of the right lung towards left hemi thorax, representing a sign of "cross ectopic lung" with left opaque hemithorax and sign of mediastinal shifting towards left side (Figures 2 a&b). CT pulmonary angiography highlighted the absence of left upper lobe pulmonary vasculature (Figure 3). Fibre optic video bronchoscopy has confirmed the blind end of the left upper lobe bronchus at the level of secondary carina shown by arrow with patent-lingular lobe bronchus (Figure 4). 2D Echo report shows mild left concentric ventricular hypertrophy. Pulmonary function test suggestive of obstructive pattern (FEV1/FVC 52.84% of predicted, FEV1 70.98% of predicted, with 23% post-bronchodilator reversibility, CBNAAT of bronchoalveolar lavage (BAL) was negative for mycobacteria. BAL for Fungal culture was also negative. The patient was managed conservatively and discharged in stable condition on oral and inhaled medication.

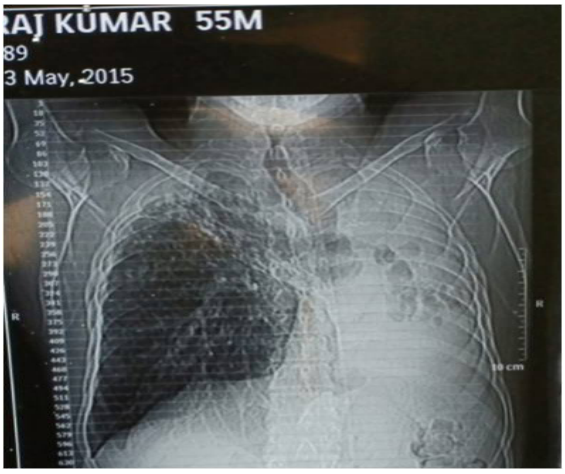

Figure 1: Computed tomography scout view of chest showing the tracheal shifting towards left hemithorax ,rising of left hemi diaphragm ,with left opaque hemithorax and compensatory hyperinflation of right lung

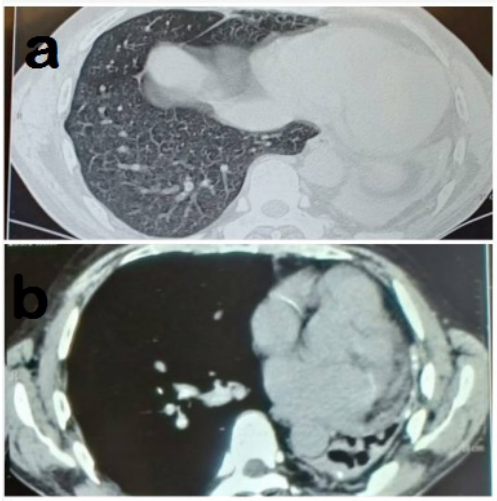

Figure 2a & b: CECT thorax in Lung window and Mediastinal window showing hyperinflation and herniation of the contralateral lung to opposite side "cross ectopic sign". volume loss of left hemithorax , compensatory hyperinflated right lung

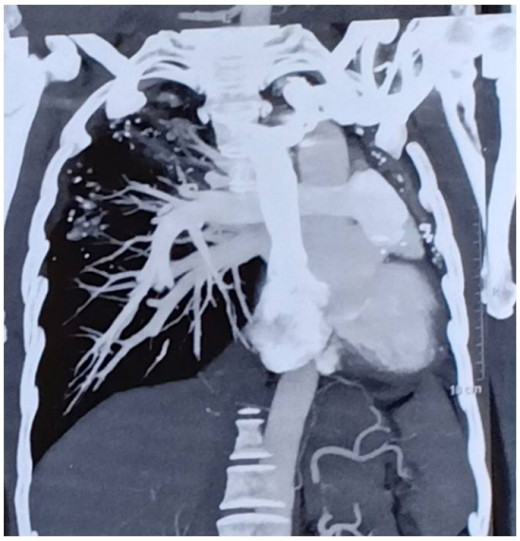

Figure 3: 3D reconstruction image of CT pulmonary angiography showed filling defect in left pulmonary artery at its origin. Unopacified tubular structure coursing superiorly closely abutting the aortic arch is seen

Discussion

Human lung development is divided into five stages: embryonic, pseudo-glandular, canalicular, saccular, and alveolar. Congenital pulmonary dysplasia developed due to defective budding of the trachea-bronchial tree sprouts from the ventral wall of the primitive foregut during 4th and 5th week of intrauterine life.[5] Laryngotracheal groove develops from the ventral wall of the pharynx and divides into the right and left lung buds at around 28th day of gestation.[12,13] Although the etiology is not very clear, genetic teratogenic agents like allopurinol, vitamin A, folic acid deficiency, or the use of salicylate during pregnancy may be responsible for it. Bilateral pulmonary hypoplasia can also appear due to

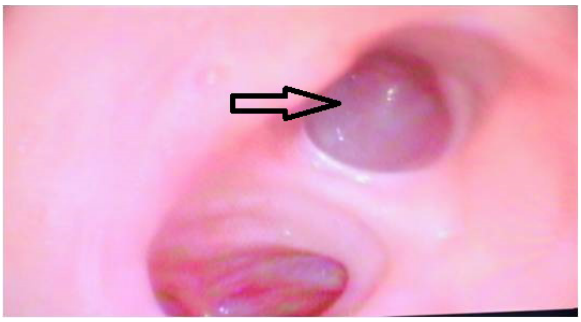

Figure 4: Fiber optic video Bronchoscopic view of obliterated left upper lobe bronchus shown by arrow with patent opening of lingular lobe

thoracic dystrophies and oligohydramnios.[8,14] Partial trisomy of chromosome 2p and 21q has also been highlighted with lung agenesis.[15] In 50% of cases, lung agenesis is complicated with other congenital anomalies like cardiovascular (nearly 1/3 of the patients have congenital heart diseases including atrial septal defect, ventricular septal defect, patent ductus arteriosus, patent foramen ovale, coarctation of the aorta or scimitar syndrome), facial (cleft lip, Mandibular hypoplasia), central nervous system, gastrointestinal (Tracheoesophageal fistulas, duodenal atresia), genitourinary system (Horseshoe kidney), skeletal system (absent rib, hemivertebrae), Down syndrome, Syndrome.[3,14] Moreover, Mardini Nyhan association is seen in consanguineous marriages, Matthew–Wood syndrome is also known as spear syndrome and micro-ophthalmia with a mutation in retinoic acid six genes (STRA6) associated with pulmonary agenesis, hydrolenthalus syndrome, Ellis Van Creveld syndrome, Opitz G/BBB syndrome, Smithlemliopitz syndrome, Meckel-syndrome, c syndrome, Fryn syndrome and Goldenhar syndrome are also linked with abnormal lung development.[12,16] Though it is absent in the reported case. In 1912, Schneider and Schwalbe classified developmental pulmonary dysplasia into three groups: Group 1 (Agenesis). Both lung and bronchus are absent; Group 2 (Aplasia): Rudimentary bronchus present but limited to blind pouch without lung parenchyma; Group3 (Hypoplasia): Hypoplastic bronchus with variable of lung parenchyma is present.[17] Later in 1955, it was modified by the Boyden into three groups according to the development of their primitive lung bud; Type1 (pulmonary agenesis): a complete absence of unilateral lung parenchyma its bronchus and vasculature, Type2: (Pulmonary aplasia): which is a complete absence of unilateral lung parenchyma and vasculature with rudimentary bronchus, Type 3 (Pulmonary Hypoplasia): Partial existence of bronchial tree with some parts of unilateral pulmonary parenchyma and its vessels.[1] our case has been classified under Type 2 Pulmonary aplasia with lobar agenesis due to the complete absence of left upper lobe parenchyma with associated vasculature through stump of left upper lobe bronchus, exists with an abrupt ending, as shown in Figure 4. Pulmonary dysplasia, especially agenesis and aplasia, are serious conditions with poor prognosis. About 50% of the children born with lung agenesis are stillborn or die within a few days due to comorbidity.[13] Mortality is higher in right lung agenesis due to tracheal compression by the aorta. However, in the present case, recurrent infection and breathlessness are the only features for frequent hospital visits. Hence, previous clinicians overlooked the diagnosis of a congenital anomaly due to the camouflage of presentation with other common manifestations like tuberculosis, pneumonia, or bronchiectasis. In adults, unilateral agenesis of the lung may mimic collapse, thickening of pleura, destroyed lung, pneumonectomy, scoliosis with pleural effusion, diaphragmatic hernia, adenomatoid cystic malformations and sequestrations.[18] Common differential diagnoses for such presentation are bronchitis, pulmonary tuberculosis, bronchiectasis, and recurrent pneumonia. However, congenital conditions like sequestration of lobe congenital cyst and congenital malformations like pulmonary dysplasia can't be denied and must be kept in mind to avoid diagnostic errors. Pulmonary aplasia is more predisposed to infection due to poor drainage of mucous secretion from rudimentary bronchus or comorbid bronchiectasis. Patients with unilateral pulmonary agenesis have also been found to have protrusion of the lung towards the opposite side that has been termed "Cross ectopic lung",[19] which is evidently highlighted in our case (Figs 2 a&b). The lungs have the ability to regenerate in children, which can lead to survival in 40 percent of cases with dysplasia or hypoplasia. Genetic and epigenetic factors influence the organogenesis of the lung. Hence, future development of gene therapy is the goal of trying to prevent lung injury which promotes lung repair to cure agenesis.[20]

Lung agenesis may remain asymptomatic rarely and could be diagnosed incidentally when a patient visits to hospital for another illness, perhaps misdiagnosed erroneously as collapsed lung or pleural effusion due to the sequel of tuberculosis in this high-burden country. With the experience of the present case, A high index of suspicion is needed for early diagnosis of lung agenesis with precise workup in patients with persistent opaque hemithorax on chest radiography to reduce the chance of diagnostic error. We had put the patient on empirical antibiotics and bronchodilators (Inhaled Corticosteroids with Long-acting beta 2 agonists) to relieve symptoms and advised regular follow-up to reduce the recurrence of infection and other associated complications. Advanced diagnostic tools like CT angiography, as well as fiber-optic video bronchoscopy, proved quite helpful in unveiling the hidden diagnosis in such a challenging case.

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