UNILATERAL PULMONARY AGENESIS: AN UNUSUAL CAUSE OF RESPIRATORY DISTRESS IN THE NEWBORN

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ABSTRACT

Unilateral pulmonary agenesis is a rare disorder and is an unusual cause of respiratory distress in the newborn. It is often associated with other congenital abnormalities, as documented in about 200 cases of unilateral pulmonary agenesis in the current literature. The onset and mode of presentation are highly variable, from asymptomatic cases discovered incidentally to symptomatic cases diagnosed in early infancy. We report a newborn infant with right pulmonary agenesis associated with facial and skeletal abnormalities who presented with respiratory distress. Unilateral pulmonary agenesis should be considered in the differential diagnosis of respiratory distress in the newborn, particularly when there are other associated congenital abnormalities.

Keywords: pulmonary agenesis, lung, abnormalities

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INTRODUCTION

Pulmonary agenesis is rare developmental defect and is defined as complete absence of the lung and its bronchus[1]. The exact incidence of this condition is not known. Autopsy studies showed an incidence of one in every 10,000 - 15,000 autopsies[1]. A prevalence of 0.0034% to 0.0097% among hospital admissions has been suggested by others[2]. Of about 200 cases of unilateral pulmonary agenesis documented in the current literature, most are associated with additional congenital abnormalities involving the skeletal, cardiovascular, gastrointestinal and genitourinary systems[3,4]. The onset and mode of presentation are remarkably variable, ranging from asymptomatic cases discovered incidentally to symptomatic cases diagnosed in early infancy. Some cases have been diagnosed in later life because of recurrent respiratory tract infections[4]. We report a case of unilateral pulmonary agenesis associated with cleft lip and alveolus and an extra pair of ribs who presented in the newborn period with respiratory distress.

CASE REPORT

A 6-day-old female infant presented with a 12-hour history of stridor, dyspnoea and cyanosis. She was born at 40 weeks gestation by normal vaginal delivery, birth weight 2.65 kg, following an uncomplicated pregnancy. Her parents were non-consanguinous. There was no family history of congenital abnormalities. Physical examination showed inspiratory stridor, cyanosis, tachypnoea with a respiratory rate of 65 /min and severe bilateral subcostal and intercostal recession. Breath sounds were markedly diminished on the right hemithorax. The heart sounds were normal and best heard over the right chest. A right unilateral complete cleft lip and alveolus were noted. The palate was normal.

Oral intubation was performed but there was difficulty in passing an endotracheal tube larger than size 2.5 mm into the trachea. Her colour improved with positive pressure ventilation. Repeat examination revealed good breath sounds on the left but inaudible on the right. Chest radiograph showed a hyperinflated left lung with mediastinal shift to the right, an opaque right hemithorax and 13 pairs of ribs (Fig 1). Echocardiography showed a normal heart displaced to the right. The provisional diagnosis on admission was gross collapse and consolidation of the right lung probably secondary to severe pneumonia.

She required assisted ventilation in the neonatal intensive care unit during the next 6 weeks. She began to deteriorate 2 weeks after admission. There were recurrent episodes of hypercapnia associated with reduced breath sounds and wheeze. She was treated with nebulized bronchodilators, aminophylline, antibiotics and steriods but failed to show any improvement. A contrast-enhanced CT scan of the chest was then performed in view of the poor response to treatment. The CT scan showed agenesis of the right lung with anterior herniation of the left lung into the right hemithorax, absent right main bronchus and left bronchial stenosis (Fig 2). She progressively deteriorated despite active medical treatment and expired on the forty-sixth day of life. Consent for a postmortem examination was not given.

Fig 1 – Chest radiograph showing an opaque right hemithorax with compensatory hyperinflation of the left lung that herniates across the midline. 13 pairs of ribs are seen.

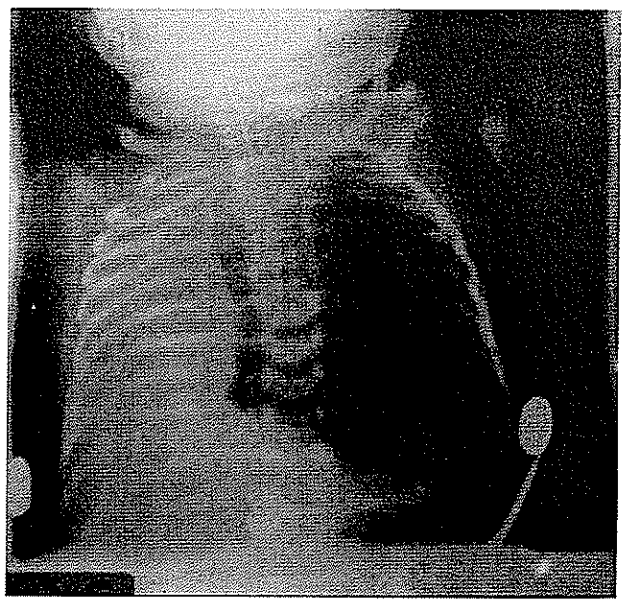

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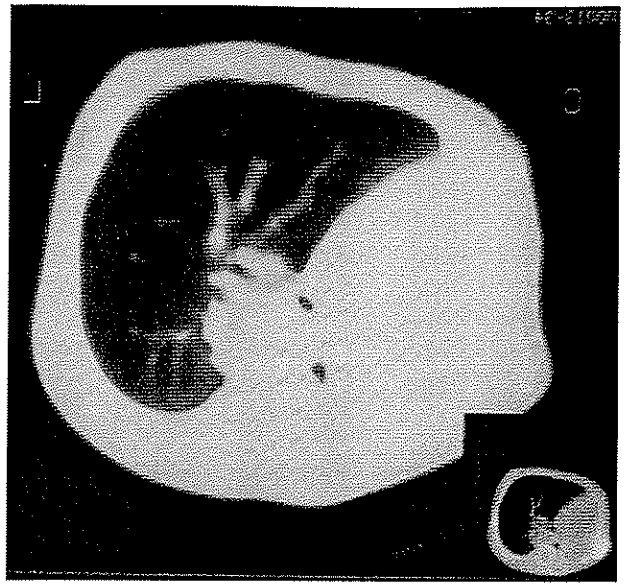
Fig 2 – Contrast-enhanced CT scan of the chest showing agenesis of the right lung with anterior herniation of the left lung.



DISCUSSION

Unilateral pulmonary agenesis is a relatively rare disorder and about 200 cases have been documented in the current literature[4]. It was first described by De Pozze who discovered it accidentally during an autopsy of an adult female in 1673[5]. Pulmonary agenesis occurs during the embryonic period when the primitive lung is forming. It can be localised to a single lobe, affect an entire lung or in rare cases, be bilateral[6]. There is no right versus left or male versus female predominance[6]. About 50% of cases have other associated congenital abnormalities[1,7]. These congenital abnormalities can affect several organ systems, including the cardiovascular, gastrointestinal, genitourinary, neurologic or musculoskeletal systems[6]. Associated congenital abnormalities in our patient include facial abnormalities in the form of cleft lip and alveolus and skeletal abnormalities with 13 pair of ribs.

Patients are usually symptomatic and present with either recurrent pulmonary infections or respiratory distress[6], as in our patient. Symptoms include dyspnoea, tachypnoea and cyanosis on exertion[6]. The time of onset of symptoms is remarkably variable[4]. However, unilateral pulmonary agenesis may be asymptomatic and discovered accidentally during routine medical examination[1]. The aetiology remains unknown[4,7]. Classical radiological features on the chest X-ray include dense homogenous opacity on the affected side, a raised hemidiaphragm, narrowed intercostal spaces and heart shadow shifted to the affected side[1,7]. Chest computer tomogram is useful in confirming the absence of pulmonary parenchyma and tracheobronchial tree[6].

Prognosis is variable and depends on the presenting symptoms and associated congenital malformations[6]. Poor prognostic factors include right-sided agenesis, recurrent infections and chronic cough[6]. Right-sided agenesis has a poorer prognosis as compared to the left because it is associated with a higher incidence of cardiac abnormalities and more severe mediastinal and cardiac displacement[2,4]. Isolated unilateral pulmonary agenesis in the absence of other congenital abnormalities can be associated with long survival and few symptoms[3,7].

Management is mostly supportive in symptomatic cases with chest physiotherapy and liberal use of antibiotics to treat any pulmonary infections[6]. Mortality and morbidity are related to associated complications and death occur due to progressive respiratory failure[4]. Half of all the reported patients die either at birth or within the first five years of life[4]. It is important to make a definitive diagnosis in asymptomatic cases who presented incidentally in order to avoid repeat investigations and irrelevant and potentially dangerous treatment such as thoracocentesis[3]. Unilateral pulmonary agenesis should be considered in symptomatic cases presenting with respiratory distress in the newborn, particularly when there are other associated congenital abnormalities as illustrated in this case report.

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