Unilateral pulmonary agenesis associated with colloidal goiter in a newborn: a case report

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Unilateral pulmonary agenesis is a very rare developmental malformation that is often associated with other anomalies. It can be asymptomatic or present with respiratory symptoms. Our case is a female newborn infant who had been taken to the hospital suffering from difficulty in breathing at the first day of birth. The baby died at the age of three days due to respiratory failure. On autopsy examination and its histopathological evaluation, we detected right pulmonary agenesis and colloidal goiter. According to the literature, pulmonary agenesis is associated with other anomalies including esophageal atresia, tracheal stenosis, musculoskeletal anomalies, DiGeorge syndrome and cardiovascular malformations such as septal defects, patent ductus arteriosus and total anomalous pulmonary venous return. To our knowledge, this is the first case of pulmonary agenesis associated with colloidal goiter.

Key words: pulmonary agenesis, colloidal goiter.

Pulmonary agenesis implies absence of a lung and its supporting vasculature, whereas the main bronchi may be either absent or hypoplastic[1]. Bilateral pulmonary agenesis is quite rare and incompatible with life. Unilateral agenesis of the lung is a more common entity; although it is often associated with other anomalies, it may be compatible with essentially normal life[2].

According to the literature, anomalies that can be associated with unilateral pulmonary agenesis are esophageal atresia, tracheoesophageal fistula, tracheal stenosis, musculoskeletal anomalies, patent ductus arteriosus and total anomalous pulmonary venous return[1,3-8]. To our knowledge, we report the first case of a female newborn infant with right pulmonary agenesis associated with colloidal goiter.

Case Report

A 2970 g female newborn was referred on her first day of life from an outside hospital because of respiratory distress. She was born by cesarean section to a 26-year-old gravida 2, para 2 mother. The mother had no significant past or obstetric history. The parents were first-degree relatives. There was no family history of any congenital anomalies. The mother did not use any drugs during her pregnancy.

On admission, the baby was cyanotic with a respiratory rate of 66/minute with chest retractions. There was absence of breath sounds in the entire right hemithorax. She was entubated and put on mechanical ventilation. Difficulty was detected while inserting the endotracheal tube towards the trachea. Her hematologic and other blood parameters were all normal. Posterior-anterior chest X-ray showed opaque right hemithorax with slight cardiac displacement, left lung hyperinflation and minimal intercostal narrowing on the right side (Fig. 1).

The baby had severe respiratory distress and her arterial blood gas analysis showed respiratory acidosis. She was treated with one dose of surfactant but no difference was detected on

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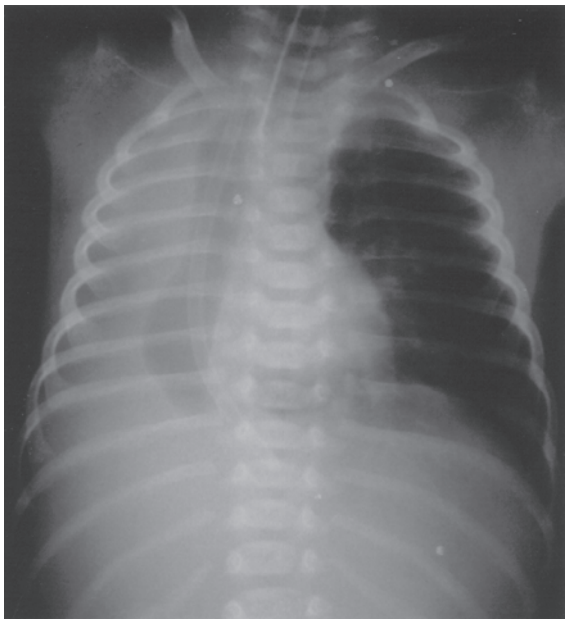

Fig. 1. Posterior-anterior chest X-ray shows opaque right hemithorax. Note the left lung. hyperinflation and slight cardiac displacement.

chest radiographs obtained after two and six hours of surfactant administration (not shown). The baby's condition deteriorated despite an increase in respiratory settings and use of vasodilators, and she died on the 38th hour of admittance.

On autopsy examination, we detected right pulmonary agenesis with its vasculature (Fig. 2). At the level of the tracheal bifurcation, there was no branching of the right main bronchus, therefore the trachea was extending directly to the left main bronchus (Fig. 3). The left lung had two lobes and was in normal range for weight. There were no other left lung, cardiac or other organ anomalies. The volume and size of the thyroid gland were increased, with a weight of 1 g and measuring 2.2 x 1.5 x 0.4 cm.

On microscopic examination, we detected histopathological findings compatible with immaturity and hypoxia, especially in the kidneys, liver and left lung. Additionally, observation of the thyroid gland showed variable size of follicles filled with colloid in incomplete nodular architecture (Fig. 4).

Our comment on the cause of death was multisystem inefficiency due to respiratory failure.

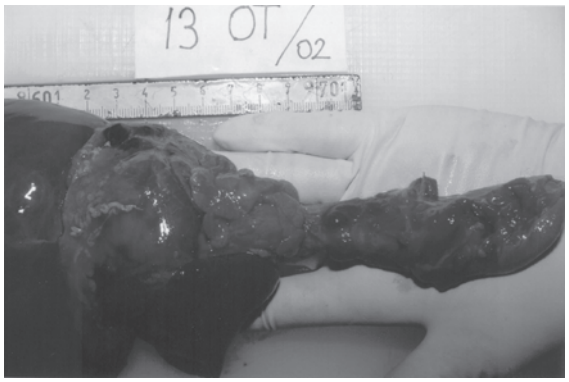

Fig. 2. Right lung agenesis, anterior aspect.

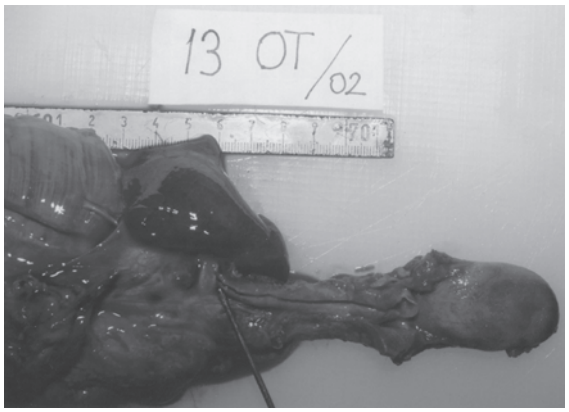

Fig. 3. On the side of the agenesis, there is no branching of main bronchus, posterior aspect.

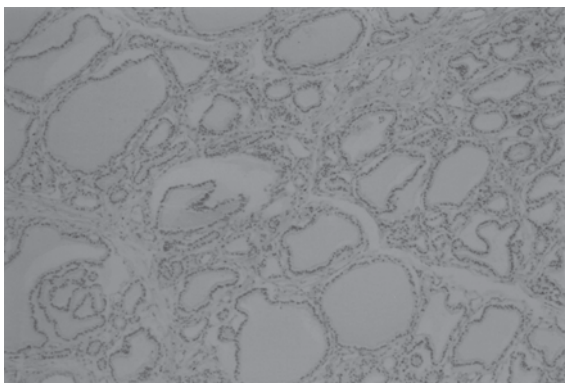

Fig. 4. Variable size of follicles filled with colloid in the thyroid gland, H&E X 100.

Discussion

The incidence of unilateral pulmonary agenesis is only one in every 10,000 to 15,000 autopsies. Congenital deficiency of the unilateral lung is believed to be caused by failure to maintain the developmental balance between the two lung buds[3].

There is a 1.3:1 female predominance with unilateral agenesis. Agenesis of the left lung appears to occur more frequently and the prognosis is generally regarded as worse for patients with agenesis on the right[2]. Our patient was female and her right lung was absent.

The larynx and upper trachea are usually normal in unilateral pulmonary agenesis. The lower trachea in unilateral pulmonary agenesis may continue directly into the existing lung as a tracheobronchus or it may bifurcate at the carina, giving rise to a rudimentary, blind-ending bronchus on the side of the agenesis.

The pulmonary artery and vein to the side of the agenesis are absent or hypoplastic[2]. In our case, there was no evidence of the vasculature and main bronchus on the side of the agenesis.

Extrapulmonary anomalies, as mentioned above, are frequently associated with pulmonary agenesis and worsen the prognosis, but even without other anomalies, children with isolated pulmonary agenesis have a shorter life expectancy than normal children, rarely surviving past their first decade.

In the newborn, colloidal goiter may result from biosynthetic defects in thyroid hormone synthesis and from maternal causes such as iodine deficiency during fetal development and antithyroid drug uptake during pregnancy. The four major biosynthetic defects in thyroid hormone synthesis are iodide transport defect, organification defect, dehalogenase defect and iodotyrosine coupling defect[9].

Our case died in her 38th hour of life because of respiratory problems. The baby had colloidal goiter in addition to right pulmonary agenesis. Her mother did not use any drugs and had no detectable endocrine organ disorders. We thus explain the etiology of the colloidal goiter as a possible biosynthetic defect in thyroid hormone synthesis.

We report the case as an incidental association. To our knowledge, it is the first such case of pulmonary agenesis associated with colloidal goiter.

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