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Unilateral Agenesis of Lung Associated with Total Anomalous Pulmonary Venous Return and Atrial Septal Defect

Case Report

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Abstract: Presented, is the case of a newborn with left-sided lung agenesis associated with total anomalous pulmonary venous return, atrial septal defect, polysplenia and hypospadias was described. Throughout the entire hospitalization period the newborn was in poor general condition, and dependent on mechanical ventilation with high oxygen concentrations. On day 20, the newborn died. According to the available literature, this case is unique in regard to the type and number of malformations.

Keywords: *Total anomalous pulmonary venous return • Atrial septal defect • Polysplenia • Hypospadias • Newborn*

1. Introduction

Congenital pulmonary anomalies are rare and can vary in their presentation and severity. Pulmonary agenesis implies absence of lungs and lung blood vessels, whereas the principal bronchus can be absent or hypoplastic [1]. This is a very rare type of malformation, which can occur isolated or associated with other anomalies such as esophageal atresia, tracheo-esophagal fistula, tracheal stenosis, cardiac malformations, musculoskeletal anomalies, anal atresia, "horse-shoe" kidney, etc. Bilateral pulmonary agenesis is condition incompatible with life [2,3].

In this paper we present a case of a newborn with left-sided lung agenesis associated with total anomalous pulmonary venous return (TAPVR), atrial septal defect (ASD), polysplenia and hypospadias. According to the available literature, this case is unique in regard to the type and number of malformations.

2. Case Report

The newborn is the sixth child from the sixth regular-course pregnancy of a 28-year old healthy mother. Gestational age was 38 weeks, Apgar score 8/9, birth weight 3.130 g. Due to global respiratory insufficiency that developed shortly after the birth, he was intubated, mechanically ventilated and treated with natural surfactant (Curosurf®Torrex Chiesi). At the 13th hour of life the child was transported to our neonatal intensive care unit (NICU).

On admission in NICU, respiratory rate was 72/min, heart rate 152/min, blood pressure 72/43 mmHg and $SatO_2$ 85%. The skin was grayish in color with pronounced central cyanosis. Auscultatory examination of the lungs revealed weak breath sounds in the right side, inaudible in the left side. Cardiac action was rhythmic, cardiac sounds were clear but unusually strong yet audible, without murmur. Glandular hypospadias was present. There were no other external anomalies.

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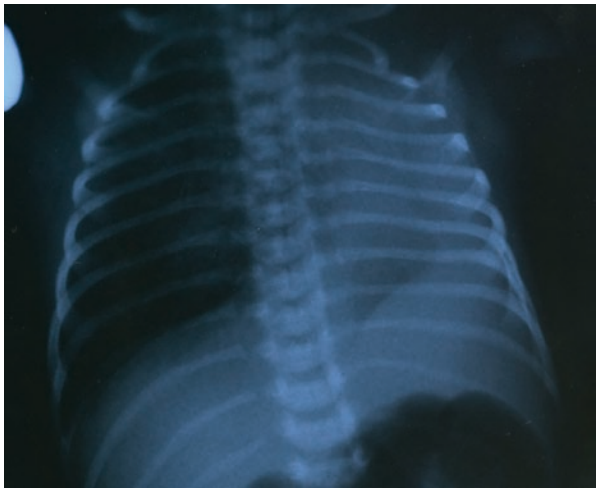
Figure 1. Radiograph revealed hyperinflation of the right lung, whereas heart shadow is completely shifted into the left.



Other than marked hypoxemia and hypercapnia, all other laboratory analyses were normal. Karyotype 46 XY.

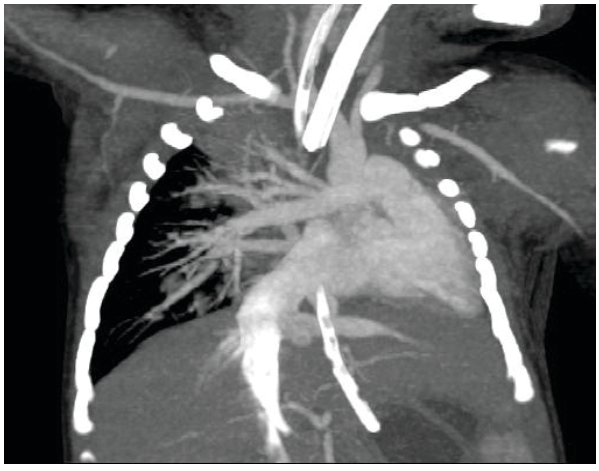
Radiography revealed hyperinflation of the right lung, whereas heart shadow is completely shifted into the left (Figure 1).

Ultrasonography findings of the brain and abdomen were normal, except for polysplenia. Echocardiography revealed patent ductus arteriosus (PDA) and foramen ovale apertum (examination was impeded due to pronounced sinistroposition of the heart).

As signs of global respiratory insufficiency persisted, high-frequency oscillatory ventilation was initiated. Chest radiography revealed total atelectasis of the left lung. As none of the applied mechanic ventilation modes resulted in any improvement, examination towards congenital lung malformation was carried out.

Bronchoscopy was performed (flexible and rigid bronchoscopy) visualizing only right principal bronchus that follows the direction of the trachea, which is shifted to the right side in the lower third, with visible estuaries of lobar bronchi. In the native and contrast computed tomography (CT) scan (Figure 2) the parenchyma of the left lung was not visualized. Multiple inflammatory condensations were present in parenchyma of the right lung. Herniation of condensed pulmonary parenchyma into the left hemithorax was visible retro-cardially. Trachea proceeds directly into the right principal bronchus. Left principal bronchus was not visible. The heart was dislocated to the left and positioned mostly in the left hemithorax; visible wide PDA 6 mm in diameter. Left lung artery was not visualized, the right lung artery was normal. Pulmonary veins on the right side formed confluence into a single draining vein, flowing into the left hepatic vein. Contrast blood was not visualized in

Figure 2. Coronal reconstruction of CT scan shows - left pulmonary artery was not visualized, the right pulmonary artery of normal morphology. Pulmonary veins on the right side form confluence into a single draining vein flowing into the left hepatic vein. Contrast blood not visualized in superior vena cava.



superior vena cava. Aortic arch was left-sided and aortic branches were normally positioned. Polysplenia was visualized. Pronounced herniation of the right lung into the left hemithorax was evident in control radiograms.

Throughout the entire hospitalization, the child was in poor general condition and dependent on mechanical ventilation with high oxygen concentrations (FiO2 0.9 to 0.97). He died 20 days after admission.

Autopsy findings confirmed left lung agenesis and associated anomalies - TAPVR, ASD, polysplenia and glandular hypospadias.

3. Discussion

Unilateral pulmonary agenesis is a relatively rare disorder and about 200 cases have been documented in the current literature. The incidence of lung agenesis has not yet been precisely determined. It is estimated to range between 0.0034-0.0097 percent among hospital admissions [1,4]. Some hypotheses suggest that abnormal development of the aortic arch during embryogenesis may possibly be the cause for development of associated anomalies [5]. In some cases an autosomal recessive inheritance was identified [6]. Spencer divided pulmonary agenesis into: 1. bilateral complete agenesis, 2. unilateral agenesis with (a) complete absence of bronchi, (b) rudimentary bronchus present but no pulmonary tissue, or (c) poorly developed main bronchus with poorly organized parenchyma, and 3. lobar agenesis [7]. Our case was consistent with the category 2a.

Left-sided lung agenesis is prevalent in almost 70% of cases, and occurs more frequently in male patients. Associated malformations occur in some 50% of cases. Most frequently, the differential diagnosis implicates atelectasis, which was also our initial hypothesis. Other diseases and conditions may reveal similar radiological pictures, such as diaphragm hernia, cystic adenomatoid malformations and sequestrations. The diagnosis is established on the basis of CT. Bronchoscopy enables direct visualization of bronchial stem, including its absence or rudimentary bronchi. Pulmonary angiography still remains the best diagnostic technique to confirm absence of ipsilateral pulmonary blood vessels [4,8].

The prognosis is highly determined by associated congenital malformations. Besides unilateral pulmonary agenesis the TAPVR (the right pulmonary veins flowed into the vena hepatica, and then into the inferior vena cava) was diagnosed in our patients, also. Finci et al. first reported the case of pulmonary agenesis associated with TAPVR [1]. The combined anomalies of pulmonary agenesis and congenital heart disease are exceedingly rare and a highly lethal association [9,10]. Clinical manifestation depends on the presence or absence of pulmonary venous duct obstructions. Obstruction of pulmonary venous return results in severe pulmonary congestion and pulmonary hypertension, indicating an urgent surgical intervention [11]. Our patient revealed distinct PDA and ASD, thus pulmonary congestion and hypertension were not pronounced.

CHD is present in some 80% of patients with polysplenia. Polysplenia may be due to multifactorial causes [12]. Polysplenia has a somewhat better prognosis than asplenia, given that its cardiac anomalies are typically less complex. In a review of 146 cases 50% died by four months and 25% survived beyond five years. Only 10% lived to adolescence [13].

To the best of our knowledge, this is the first reported case of a newborn with left-sided pulmonary agenesis associated with total anomalous pulmonary venous return, atrial septal defect, polysplenia and hypospadias.

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