Unilateral lung agenesis – rare congenital malformation diagnosed in infants

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ABSTRACT

Pulmonary agenesis is a congenital anomaly defined by the complete absence of lung parenchyma, as well as bronchial and vascular structures. Right lung agenesis is less frequent compared to left lung agenesis, and usually has a worse prognosis being associated with other congenital anomalies. The study focuses on the clinical case of a child with left lung agenesis.

Keywords: left pulmonary agenesis, pulmonary agenesis, children

INTRODUCTION

Congenital lung malformations comprise a heterogeneous group of developmental abnormalities. Most of these malformations are detected in early childhood. Only a very small number remains asymptomatic and undiagnosed until adulthood. In these cases, the detection of the malformation happens accidentally when a chest X-ray is taken [1].

Pulmonary agenesis is an unusual congenital anomaly, which is defined by the complete absence of lung parenchyma, as well as bronchial and vascular structures, which develops when there is an interruption in the evolution of the primitive lung bud. Right pulmonary agenesis is less frequent compared to left pulmonary agenesis, which occurs in approximately 70% of cases, and usually has a worse prognosis, being associated with other congenital anomalies [2,3].

According to data from the literature, pulmonary agenesis has a prevalence of 34 cases per 1,000,000 live births and 1 case per 10,000-15,000 autopsies with predominance in the female gender [4].

The etiology of the malformation is unknown, but the pathogenetic mechanisms may be associated with genetic errors in the reproduction of the distal part of the arm of chromosomes 2, with etiologies caused by viruses or vitamin A deficiency [5].

In children with pulmonary agenesis, the first clinical manifestations that predominate are cyanosis and respiratory disorders, dyspnea, which appear since the neonatal period. During the physical examination, the asymmetry of the ribcage and the propagation, the accentuation of the heart sounds in the hemithorax where there is no lung can be detected [6].

The asymmetry of the chest is characterized by the reduction in the size of the chest with a predominance of the reduction of the inspiratory amplitude, thus children can often present scoliosis. In these patients, the displacement of the mediastinum, heart and large vessels towards the affected hemithorax prevails, with the reduction of the intercostal space and the elevation of the diaphragm [7].

To confirm the diagnosis, a chest x-ray is performed where a small, completely opaque ipsilateral hemithorax is found. In most cases of unilateral pulmonary agenesis, the mediastinum herniates and moves in the ipsilateral direction, this can be confused with partial atelectasis or pneumonic consolidation of the ipsilateral lung, so CT and MRI are of great value for a definite diagnosis [8].
The treatment of children with pulmonary agenesis is based on the associated pathologies, antibiotic therapy in the repeated lung infections they manifest [6].

Although certain hypotheses or risk factors are mentioned in some studies, the etiology of this malformation remains unknown. In most cases, patients with pulmonary agenesis are associated with other malformations, especially cardiovascular, gastrointestinal, genito-urinary and musculoskeletal, which worsen the prognosis, thus determining a mortality rate of up to 33% in the first year of life and up to 50% in the first 5 years [9].

**CLINICAL CASE PRESENTATION**

**History of the disease.** The child is considered sick since 05.12.2023 with allegations of moderate dyspnea, dry cough. He is redirected to the Mother and Child Institute with boarding in the RTI section, later in the pneumology section for treatment and examination.

**Life history.** The child is from pregnancy III, which evolved without particularities. During the pregnancy, an USG examination was performed at the 7-month term, which did not show any suspicion of pulmonary malformation. Child from birth III, physiological at 39 weeks, birth weight – 2450 g, waist – 49 cm. Vaccinated in maternity (HBV, BCG).

**Objective examination.** General condition severe-medium with dry cough, rhinorrhea, severe dyspnea on exertion, moderate substernal draft, lack of appetite, apathy. FR – 32 resp./min, FCC – 125 beats/min, t – 36.6°C, SpO₂ – 98%. Pale skin, presence of hemangiomas in the right retroauricular region and on the chest (Figure 1).

Subcutaneous adipose tissue insufficiently developed, weight 6100 g (PI=0.8), weight deficit gr. I. Satisfactory muscle development, normal muscle strength. Osteo-articular system: normal head shape, unilateral flattening of the ribcage, moderate substernal draft is present. Subduality in the projection of the left lung, auscultation of rough breathing, wet, crepitated rales are perceived on the right, and in the left hemithorax the breathing is diminished basally, medially.

**Cardiovascular system.** The boundaries of the heart are shifted to the left side, the heart sounds are rhythmic, sonorous, they propagate on the projection area of the left lung.

**Digestive system.** Mucous membranes of the oral cavity moist, slightly hyperemic pharyngeal isthmus, lack of teeth. The abdomen is soft, tender to palpation, tympanic sound, the liver +1 cm from the right costal edge.

**Renal-urinary system:** absent edema, kidneys and bladder are not palpable, free urination.

**Blood count.** Hemoglobin 125 g/l; erythrocytes \(4.46 \times 10^6/\mu L\), hematocrit 40%, leukocytes \(8.8 \times 10^9/L\), neutrophils 31.2%, eosinophils 6.9%, basophils 0.6%, lymphocytes 53.5%, monocytes 7.8%, ESR 3 mm/h.

**Sputum bacteriology.** *Klebsiella pneumoniae* titer \(10^5\) resistant to amoxicillin, cefazolin, cefuroxi-
me, cefepime, cefoperazone, cefotaxime, ceftiraxone and sensitive to ciprofloxacin, levofloxacin, co-trimoxazole, meropenem.

**Serum biochemistry.** Alanine aminotransferase 20.00 U/L; Aspartate aminotransferase 30.70 U/L; Calcium 2.63 mmol/l; Serum creatinine 34.00 µmol/l; Creatine kinase 200.00 U/L; Creatine kinase MB 264 U/L; Cardiac troponin 0.33 0 - 1.68 · ng/ml; Glucose 4.80 µmol/l; Lactate dehydrogenase 299.00 U/L; Potassium 4.20 mmol/l; Sodium 135.00 mmol/l; Urea 2.90; D-Dimer 0.34 mg/l; Procalcitonin 0.30 ng/ml.

**EKG.** Irregular sinus rhythm 133. AE-SI Q3 (vertical cord). RV hypertrophy. Right bundle branch block of Fas.HIs. Disorders of repolarization processes with a diffuse character.

**Echo cord.** The regional contraction function of the LV myocardium is not affected. Fluid in the pericardial cavity after LV 2.0 mm and after RV-4.0 mm. The cavities of the heart are not dilated. The pump function of the LV myocardium – within the limits of the norm. False cord in VS cavity. Tricuspid vein insufficiency grade I. Pulmonary vein insufficiency grade I. Mild HP. PSAP 37 mmHg. VS cavity (Dd=27mm). The pump function of the LV myocardium within the limits of the norm (FE=67%).

**Pulmonary USG.** In the pleural cavity on the left small liquid collection: anterior >9 mm, posterior 30×11 mm.

**Computed tomography.** Is visualized a single expanded right lung with contralateral herniation towards the left hemithorax (Figure 2), the single right pulmonary artery with a diameter of 9.4 mm (Figure 3) and an aberrant vessel arising from the pulmonary trunk, the posterior portion with a diameter of 2.1 mm (Figure 4). Conclusion: CT imaging data suggestive of left-sided pulmonary agenesis. Pneumonic infiltration with apical atelecatic component on the right. Mucus deposits at the level of the upper right main bronchus.


**FIGURE 2.** Left-sided pulmonary agenesis
DISCUSSIONS

By 2022, 259 cases have been identified, of which 59%-right pulmonary agenesis, 34%-left pulmonary agenesis and 7%-bilateral agenesis [10].

Scientific findings reveal the fact that patients with right lung agenesis are more frequently associated with other malformations compared to patients with left lung agenesis, referring to our patient, we have disorders only at the level of the...

FIGURE 3. Single pulmonary artery on the right

FIGURE 4. Aberrant vessel emerging from the pulmonary trunk
cardiovascular system, which imply a better prognosis of viability favorable [11].

As far as our patient’s case is concerned, a qualitative management is necessary to prevent solitary lung infections, or treatment as early as possible so as not to endanger the child’s life. Asymptomatic cases or patients with minimal symptoms have a good prognosis for life [3].

CONCLUSIONS

1. In the absence of other malformations, unilateral pulmonary agenesis is compatible with life, but is often associated with severe respiratory infections during childhood, which are associated with high mortality rates.

2. An important goal in the detection of pulmonary malformation is to establish a correct diagnosis from the intrauterine period.

3. The child with pulmonary malformations requires extensive surveillance in dynamics with the recording of parameters: FR, FCC, SaO2, t°C, BP, and monitoring by a multidisciplinary team, consisting of pneumologist, cardiologist, thoracic surgeon, radiologist, geneticist, neurologist.

4. Life prognosis is reserved due to serious complications, which emphasizes the dynamic importance of children with pulmonary malformation, also associated with other anomalies such as cardiovascular ones present in the patient in the exposed case.

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REFERENCES


