

Unilateral lung agenesis – rare congenital malformation diagnosed in infants

By Svetlana Sciuca

Unilateral lung agenesis – rare congenital malformation diagnosed in infants

Ala David¹, Rodica Selevestru², Iana Coropceanu², Carolina Mighic², Eva Gudumac²,
Svetlana Sciuca²

¹Medicover, Romania

⁸

²“Nicolae Testemitanu” State University of Medicine and Pharmacy, Republic of
Moldova

¹ 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.

Next, the clinical case of a child with left lung agenesis will be presented.

Key words: left pulmonary agenesis, pulmonary agenesis, children

⁴
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 [2].

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,

Svetlana Sciuca
svetlana.sciuca@usmf.md

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 [4,11].

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 [7].

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 [1].

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 [5].

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 [3].

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 [6].

The treatment of children with pulmonary agenesis is based on the associated pathologies, antibiotic therapy in the repeated lung infections they manifest [5].

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 subcostal draft, lack of appetite, apathy. FR – 32 resp./min, FCC – 125 beats/min, t – 36.6°C, SpO2 – 98%. Pale skin,



(a)



(b)

presence of hemangiomas in the right retroauricular region and on the chest (fig.1)

Figure 1. Hemangiomas in the right retroauricular region (a) and on the chest (b)

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 subcostal 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 $4.46 \times 10^6/\mu\text{L}$, hematocrit 40%, leukocytes $8.8 \times 10^9/\text{L}$, 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, cefuroxime, cefepime, cefoperazone, cefotaxime, ceftriaxone 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 $\mu\text{mol/l}$; Creatine kinase 200.00 U/L; Creatine kinase MB 264 U/L; Cardiac troponin 0.33 0 - 1.68 · ng/ml; Glucose 4.80 $\mu\text{mol/l}$; 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 30x11 mm.

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

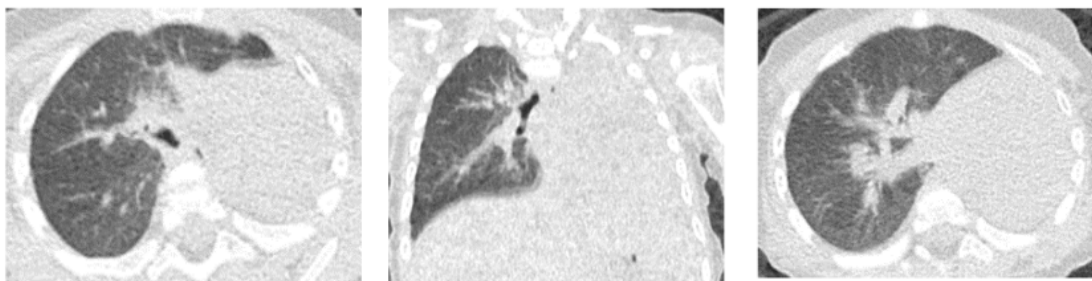


Figure 2. Left-sided pulmonary agenesis

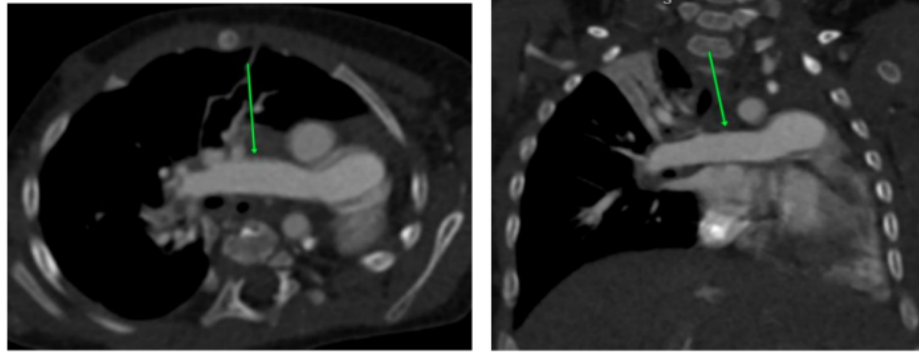


Figure 3. Single pulmonary artery on the right

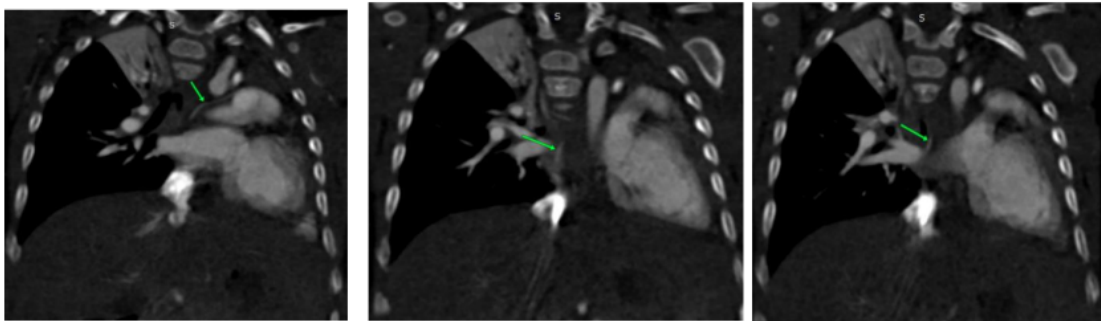


Figure 4. Aberrant vessel emerging from the pulmonary trunk

Clinical diagnosis. (J150) Pneumonia due to *Klebsiella pneumoniae* on the right, moderate form. (J960) Acute respiratory failure gr I. (I301) Post inflammatory acute infectious pericarditis. (I500) Congestive heart failure IC I CF NIHA. (E46) Protein-energy malnutrition, unspecified. (Q333) Agenesis of the left lung.

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 cardiovascular system, which imply a better prognosis of viability favorable [8].

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. ⁹ Asymptomatic cases or patients with minimal symptoms have a good prognosis for life [11].

Conclusions:

- ² 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 cardiac vascular ones present in the patient in the exposed case.

Bibliography:

1. Bourama Kané, Mody Abdoulaye Camara, Seydou Togo, Mohamed Maba Traoré, Boubacar Mami Touré, Moussa Abdoulaye Ouattara. Isolated Right Pulmonary Agenesis in a 2-Month-Old Infant in the Pediatric Ward of Hospital of Mali in *Open Journal of Pediatrics*, 2020.
2. Diana Alexandra Pimenta, corresponding author Filipa Lemos Aguiar, Beatriz Celeiros Fernandes, and Rui Rolo. Late diagnosis of pulmonary agenesis in *BMJ Case Reports*, 2021.
3. Edward A. Boyden Ph.D. Developmental anomalies of the lungs in *The American Journal of Surgery*, pp. 79-89, 1955.
4. Elias Gonçalves, Ofélia Sachicola, Bartolomeu Estanislau, Francisca Quifica, Humberto Morais, Margarete Arrais. Agenesis of the right lung in an adult woman: A case report in *Clinical Case Reports*, 2023.
5. Gabriel Aprodu. *Chirurgie pediatrică Ediția a III-a revizuită și adăugită* pp.40-45, 2013.
6. Ji Young Kim, Woo Sun Kim, corresponding author Kyung Soo Lee, Bo-Kyung Je, Ji Eun Park, Young Jin Ryu, Young Hun Choi, and Jung-Eun Cheon. Posterior Lung Herniation in Pulmonary Agenesis and Aplasia: Chest Radiograph and Cross-Sectional Imaging Correlation in *Korean Journal of Radiology*, 2021.
7. Paula Vanessa Valverde Dinamarca and Cesar Cilento Ponce. Pulmonary agenesis and respiratory failure in childhood in *Autopsy Case Reports*, 2015.
8. Russell, Bronwyn C BS., Whitecar, Paul MD, Nitsche, Joshua F. MD, PhD. Isolated Unilateral Pulmonary Agenesis and Other Fetal Thoracic Anomalies in *Obstetrical & Gynecological Survey*, 2014.
9. S. Aimée Kissou, Souleymane B. W. Adjaba, Jacqueline Tamini, and Hélène Traore. Unilateral Pulmonary Agenesis about a Case Revealed by an Acute Respiratory Infection in a Young Infant in *Case Reports in Pediatrics*, 2023.
10. Shoji Fukuoka MD, Kenichiro Yamamura MD, PhD, MSc, Hazumu Nagata MD, PhD, Daisuke Toyomura MD, Yusaku Nagatomo MD, PhD, Yoshimi Eguchi MD, Kiyoshi Uike MD, PhD, Yuichiro Hirata MD, Hirotsuke Inoue MD, PhD, Masayuki

Ochiai MD, PhD, Shouichi Ohga MD, PhD. Clinical outcomes of pulmonary agenesis: A systematic review of the literature in *Pediatric Pulmonology*, 2022.

11. Urvinderpal Singh, Daksh Jhim, Sunil Kumar, Vidhu Mittal, Navdeep Singh, Hitesh Gour and Muralidharan Ramaraj. Unilateral Agenesis of the Lung: A Rare Entity in *Am J Case Reports*, 2015.