ramr
American Review of Respiratory Medicine
Revista Americana de Medicina Respiratoria

CASE REPORT

ISSN 1852-236X

Congenital Pulmonary Airway Malformation in a Newborn: Case Report

Malformación congénita de la vía aérea pulmonar en un recién nacido: Reporte de caso

Stand Niño Iván[1]; Restrepo Hincapié Andrea[2]; Aristizábal Ardila Paula[3]; Sánchez Silvera Zaira[4]; Peña Soto Jorge[4]; Ovalle Villadiego Carlota[5]

Received: 04/15/2025
Accepted: 09/16/2025

Correspondence
Andrea Restrepo Hincapié.
E-mail: andreacrisrh@gmail.com

ABSTRACT

Congenital lung malformations represent a disruption both in the formation of functional pulmonary parenchyma and in the distribution of pulmonary vasculature. Prenatal imaging studies have become widely accessible tools for detection of congenital disorders, contributing to increase the incidence of this condition. However, the classification, diagnosis, and treatment of this disorder are still a challenge in clinical practice. We present the case of a female newborn with a prenatal history of congenital pulmonary airway malformation, initially detected through routine obstetric ultrasound and confirmed postnatally by computed tomography and histopathological analysis. This case report aims to describe a disease with a complex classification and variable clinical course.

Key words: Lung; Congenital lung malformation; Lung volume reduction surgery

RESUMEN

Las malformaciones pulmonares congénitas constituyen un factor disruptivo tanto en la formación de parénquima pulmonar funcional, como en la distribución de los vasos sanguíneos. Los estudios de imagen en la etapa gestacional se han convertido en una herramienta ampliamente disponible para la detección de patologías congénitas, que a su vez han contribuido a un aumento en la incidencia de esta entidad. Sin embargo en la práctica clínica, sigue siendo un reto la nomenclatura, el diagnóstico y el tratamiento de este trastorno. Presentamos el caso de un recién nacido de sexo femenino con antecedente prenatal de malformación congénita de la vía aérea pulmonar evidenciado en ultrasonido de rutina y confirmado en el periodo posnatal por tomografía y estudio histopatológico. Este reporte de caso tiene como objetivo describir una entidad de clasificación compleja con un curso clínico variable.

Palabras clave: Pulmón; Malformación congénita pulmonar; Cirugía de reducción de volumen pulmonar.

Rev Am Med Resp 2025;25:145-150. https://doi.org/10.56538/ramr.AZIR8143

[1] Pediatric Pulmonologist, Escuela Colombiana de Medicina, Bogotá – Colombia. Postgraduate Pediatrics Professor, Barranquilla - Colombia.
[2] Pediatrics Resident, Universidad Libre, Barranquilla - Colombia.
[3] Pediatrics Resident, Universidad Simón Bolívar, Barranquilla - Colombia.
[4] Pediatrics Resident, Universidad Metropolitana, Barranquilla - Colombia.
[5] Pediatrics Resident, Universidad del Norte, Barranquilla - Colombia.

INTRODUCTION

Congenital lung malformations are a group of diseases with a prevalence of 30 to 42 cases per 100,000 people, and in some studies they represent between 5% and 18% of all congenital anomalies. In recent years, the incidence of this condition has increased thanks to advances in ultrasound technology for screening fetal malformations, so in the long term it is expected to be excluded from the list of rare disorders.[1]

The current nomenclature remains complex, as it takes into account criteria such as genetic variants, morphological lesions, histological patterns, and clinical manifestations –among other factors– that are useful for practical differentiation but are not mutually exclusive. Langston's classification describes several pathological findings: large-cyst cystic adenomatoid malformation (Stocker 1), small-cyst cystic adenomatoid malformation (Stocker 2), solid/adenomatoid cystic adenomatoid malformation (Stocker 3); extralobar sequestration; bronchogenic cyst; congenital lobar hyperinflation; pulmonary hyperplasia; bronchial atresia. In 2002, five subtypes of congenital pulmonary airway malformation were included, corresponding to a new classification proposed by Stocker, based on anomalies that occur at different levels of the tracheobronchial tree and at different stages of lung development.[2,3]

The diagnostic method of choice in the prenatal period is ultrasound, as it offers a good safety profile and is considered a reproducible and cost-effective study. In the postnatal evaluation, chest computed tomography is the preferred study, offering superior diagnostic performance compared with other modalities. Low concordance has been demonstrated between prenatal ultrasound and postnatal histological examination, making ultrasound insufficient as the sole diagnostic method, whereas postnatal chest CT shows greater concordance with histological findings, especially in the detection of abnormal systemic vessels.[4]

Expectant management is an option for asymptomatic patients with lesions of favorable prognosis. However, in cases with hemodynamic instability or high risk for the later development of infections or neoplasms, anatomical resection is the recommended treatment.[5]

CASE DESCRIPTION

A female newborn, the product of a second pregnancy that was adequately monitored, born to a 21-year-old mother with no comorbidities. An obstetrical ultrasound performed at 24 weeks of gestation described increased bilateral pulmonary echogenicity accompanied by displacement of the cardiac axis to the right, with no other identifiable fetal morphological abnormalities. This ultrasound study was concluded as consistent with cystic adenomatoid malformation of the lung. Delivery occurred by cesarean section at 36 weeks of gestation due to the onset of preterm labor and the previously described fetal comorbidity. She was born weighing 2,900 grams, measuring 49 centimeters in length, with a head circumference of 33 centimeters, and had a low Apgar score of 4/10, for which she required high-frequency invasive mechanical ventilation as well as inotropic and inodilator support. On head-to-toe examination, a significant finding was a markedly decreased vesicular breath sound over the left hemithorax. Postnatal chest radiography and computed tomography showed extensive involvement of the left lung parenchyma by cystic lesions producing a mass effect, displacing mediastinal structures, with significant atelectasis of the ipsilateral lower lobe, in addition to small cystic foci in the upper and lower lobes of the right lung (Figure 1). Echocardiography revealed dextrocardia with situs solitus, tricuspid regurgitation, mitral insufficiency, and pulmonary hypertension. At 8 days of life, she underwent a surgical procedure through video-assisted thoracoscopy, which revealed a large cystic mass involving both lobes of the left lung, especially the upper lobe. A left upper lobectomy via thoracotomy was required due to the challenging approach posed by the lesion's large size. The specimen was studied by the Pathology Department, which described a rectangular, blackish-colored mass measuring 7 × 5 × 3 cm, with a firm consistency but with softened areas. Microscopically, adenomatous and cystic regions delimited by simple cuboidal to low columnar epithelium were observed, accompanied by recent intra-alveolar hemorrhages and no evidence of neoplastic infiltration (Figure 2). Based on the clinical, histological, and imaging findings, a diagnosis of congenital pulmonary airway malformation type III was made. After surgery, her postoperative

course was poor, with no possibility of ventilator weaning and an increased need for vasoactive support; she therefore died at 29 days of life.

DISCUSSION

Congenital pulmonary airway malformation (CPAM) is defined as a disorder of the airways secondary to an alteration of the epithelial component of the bronchial bud, which manifests as exaggerated growth of bronchial structures and a reduction in the number of alveoli. This predisposes to the development of a multicystic mass of nonfunctioning lung tissue and/or adenomatous pulmonary areas. The inheritance pattern is usually sporadic, and to date no aggravating maternal factors have been described.[6,7]

The nomenclature has changed since 1949, when it was first described by Ch'in and Tang as congenital cystic adenomatoid malformation (CCAM). However, it was later shown that cystic lesions were present in only three of the five subtypes described by Stocker, and adenomatous lesions in only one subtype, which could lead to confusion when categorizing them. In 1977, Stocker replaced the term CCAM with CPAM, as it is currently known, and grouped the lesions into three types. In 2002, the same author expanded this classification to five subtypes.[3,5]

Type 0 lesions, also known as tracheobronchial lesions or acinar dysplasia, are the least frequent, accounting for 1–3% of all subtypes. Involvement is bilateral, with small, hypoplastic lungs, and this condition is considered incompatible with life. Type 1 lesions, or bronchial/bronchiolar lesions, are the most frequent, accounting for 60–65% of cases. They are usually unilobar and may be morphologically multicystic or present with a dominant cyst. Patients with this abnormality may have respiratory symptoms, but prognosis improves with surgical resection of the affected anatomical segment. Type 2 lesions, or bronchiolar

Figure 1. Panel A: Chest X-ray showing displacement of mediastinal structures due to cystic lesions. Panel B: Chest CT scan (lung window) showing, on axial view, multiple cysts in the left hemithorax.

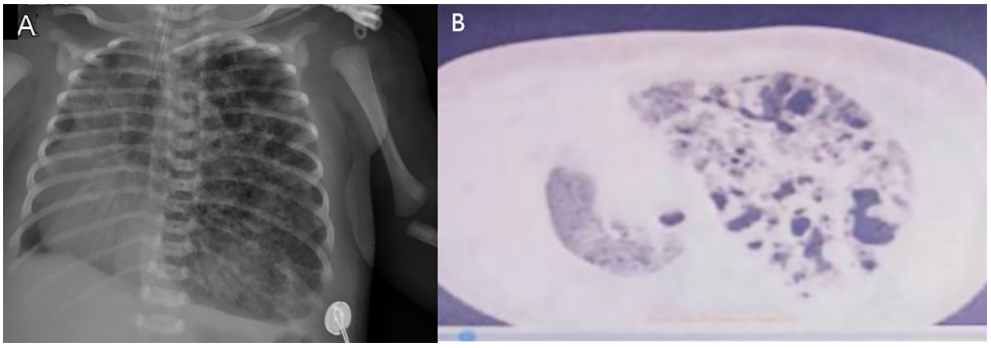
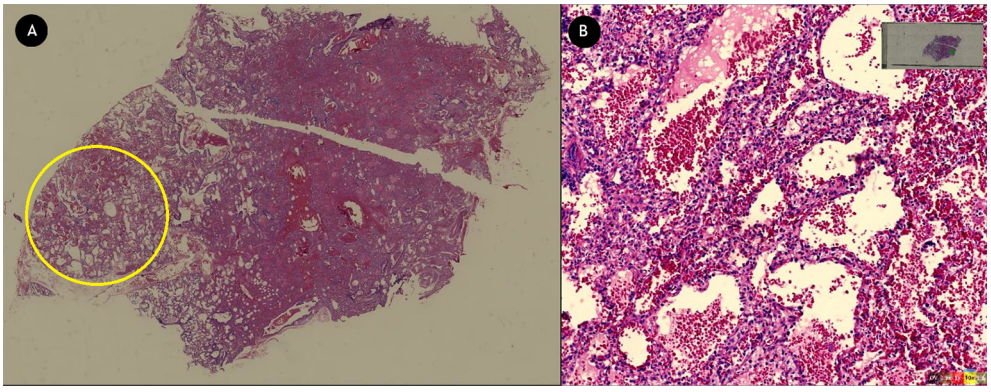


Figure 2. Panel A: Histological section of lung tissue stained with H&E. (Panoramic view). An area of tissue replacing normal alveolar tissue is outlined, forming a multicystic pattern. Panel B: Microscopic detail of the structures showing loss of the normal alveolar pattern associated with intraalveolar hemorrhages.



lesions, are usually diagnosed in the first year of life. They may partially involve a lobe or, in some cases, an entire lung. This subtype has been associated with other congenital malformations in up to 50% of cases. Type 3 lesions, or bronchiolar/alveolar lesions, are usually diagnosed in utero due to the presence of a large adenomatous-like mass that generally involves an entire lung. These patients are born with severe respiratory distress due to mediastinal displacement. Type 4 lesions, or alveolar lesions, are hamartomatous malformations of the acinus. They tend to involve a single lobe, are often diagnosed incidentally due to pneumothorax or infectious processes, and are usually associated with neoplasms. [5, 8]

Considering recent advances in terms of availability of prenatal diagnostic methods, Andrew Bush, one of the most widely recognized pediatric pulmonologists today, points out the inconsistency that may arise when using the term congenital cystic adenomatoid malformation in both the prenatal and postnatal periods. In the prenatal stage, this term may be applied to a lesion that could even disappear before birth; however, in the postnatal period, it is also used to describe an anomaly severe enough to require a lobectomy. Therefore, to achieve greater diagnostic accuracy, it is suggested to use clear, descriptive language for findings, avoiding speculation on embryological origins and keeping clinical and pathological descriptions separate. [9]

At present, the exact cellular mechanisms involved in the pathogenesis of this disorder remain under investigation. Studies in transgenic mice have identified the overexpression of fibroblast growth factors (FGF) types 7 and 10 in the pulmonary mesenchyme, which interfere with lung morphogenesis in CPAM. Acinar dysplasia has been associated with genes encoding the transcription factor TBX4, in which there is a disruption of the TBX4–FGF epithelial–mesenchymal signaling pathway. In type 1 lesions, atypical hyperplasia of goblet cells has been described, which may predispose to mucinous adenocarcinoma. The distinction between type 1 and type 2 lesions is complex, since mutations have been reported in both, in the KRAS, GNAS, and EGFR genes. Similarly, it has been suggested that type 3 lesions may result from mosaic KRAS mutations arising in the pulmonary epithelium during early stages of development, placing them within the growing group of RASopathies; however, no association between bronchiolar/alveolar lesions and mucinous adenocarcinoma has been reported. Type 4 lesions have been studied in several families, and in some cases high-grade rhabdomyosarcomatous features have been identified. In this subtype, mutations in the DICER1 gene have been detected in more than 70% of children with alveolar lesions, in addition to others identified in the TP53 gene. Recent studies have suggested a change in the designation of this subtype, considering it to be a pleuropulmonary blastoma.[10,11]

The treatment of choice for congenital pulmonary malformations with poor prognosis and significant clinical impact at an early age remains surgical; however, in asymptomatic patients with a high life expectancy, this option is debatable. Although pulmonary resection is considered an alternative to preserve alveolar capacity, minimize the risk of malignancy, and prevent infectious complications, the risk of subsequent respiratory morbidity must be taken into account. Expectant management is a reasonable option for small lesions accompanied by mild symptoms, and a chest CT scan is suggested around 6 months of age, according to each patient's particular circumstances, since lesions detected prenatally have been described to regress spontaneously during postnatal evolution.[12]

Long-term follow-up of post-surgical patients with congenital pulmonary malformation is very limited. In a study conducted in Spain, acute postoperative complications were reported in 28% of the studied population; however, follow-up was performed only during the first month after pulmonary lobectomy. In Italy, patients undergoing pulmonary resection were assessed, with long-term effects after one year of follow-up ranked by frequency as follows: chronic cough, recurrent infections, wheezing, poor exercise tolerance, and the development of chest and spinal deformities. These findings were not statistically significant for most of the variables studied, except for spirometry results, in which a significant correlation was demonstrated between pneumonectomy and long-term deterioration of pulmonary function. Further follow-up studies are needed to explain the risk–benefit ratio of surgical procedures in patients with CPAM, considering that in the previously described single-center studies, this was the most frequent diagnosis among congenital pulmonary

malformations and that most patients remained free of respiratory symptoms prior to surgical intervention.[13,14]

In Latin America, some descriptive studies of patients with congenital pulmonary malformations have been conducted, with CPAM being the most common type of lesion, followed by pulmonary sequestration and hybrid lesions. Regarding the clinical spectrum, and in contrast to European countries, a higher proportion of patients have been found to be symptomatic prior to the procedure, ranging from potentially fatal perinatal respiratory failure to recurrent bronchopulmonary infections in older children.[15,16]

In this case report, the patient was born with a large pulmonary lesion with respiratory and hemodynamic compromise; therefore, the final outcome was death, despite timely surgical treatment. The changes reported on echocardiography may be secondary to displacement of mediastinal structures; however, the possibility that these findings indicate associated congenital malformations cannot be ruled out.

CONCLUSION

Congenital pulmonary malformations represent a heterogeneous group of disorders that require the participation of multiple specialties in order to harmonize therapeutic approaches. The dilemma between conservative management and surgical treatment remains controversial in patients with mild symptoms; therefore, more prospective studies are needed.

ETHICAL CONSIDERATIONS

This clinical case was previously presented at the XII Symposium on Rare Diseases, held in March 2025 in the city of Barranquilla, where it was presented in poster format. This article is not currently under consideration by another journal. Permission was obtained from the individuals responsible for the child's care to collect the information, and confidentiality was guaranteed; all of this was clearly explained verbally and supported through informed consent. This manuscript has been read and approved by all authors listed in the publication, who also declare that they have no potential conflicts of interest. This research did not receive funding, and no experiments involving humans or animals apply in this case.

Conflict of interest

The authors have no conflict of interest to declare.

REFERENCES

1. Cancemi G, Distefano G, Vitaliti G, et al. Congenital Lung Malformations: A Pictorial Review of Imaging Findings and a Practical Guide for Diagnosis. Children (Basel). 2024;25;11:638. https//doi.org/10.3390/children11060638.
2. Lagston C. New concepts in the pathology of congenital lung malformations. Semin Pediatr Surg 2003;12:17-37. https//doi.org/10.1053/spsu.2003.00001.
3. Stocker J. Congenital pulmonary airway malformation - a new name for and an expanded classification of congenital cystic adenomatoid malformation of the lung. Histopathology 2002;41(Suppl.2):424-58. https://www.researchgate.net/publication/284194320/
4. López-Díaz M, Antón-Pacheco JL, Gallego-Herrero C, et al. Diagnostic accuracy of imaging compared to histology in congenital lung malformations. An Pediatr (Engl Ed). 2023;99:304-11. https://doi.org/10.1016/j.anpede.2023.10.002.
5. Saavedra M, Guelfand M. Enfoque actual de las malformaciones pulmonares. Revista médica clínica Las Condes. 2017;28:29-36. https://doi.org/10.1016/j.rmclc.2017.01.003.
6. Macías Zambrano F, Garzón Ávila H, Vela Merino D, Santamaría A, Herdoíza Arroyo, L. Malformación congénita de la vía aérea pulmonar. Reporte de un caso. Metro Ciencia, 2020;28;21-8. https://doi.org/10.47464/MetroCiencia/vol28/4/2020/21-28
7. Mehta PA, Sharma G. Malformación congénita de las vías respiratorias pulmonares. [Actualizado el 7 de agosto de 2023]. En: StatPearls [Internet]. Treasure Island (FL): StatPearls. https://www.ncbi.nlm.nih.gov/books/NBK551664/
8. Valmaggia C, Guadalupe A, Machado K. Malformación congénita de la vía aérea pulmonar: a propósito de un caso clínico. Arch Pediat Urug 2022;3(2):e309. https://doi.org/10.31134/ap.93.2.25
9. Bush A. Congenital lung disease: a plea for clear thinking and clear nomenclature. Pediatr Pulmonol. 2001;32:328-37. https://doi.org/10.1002/ppul.1126
10. Pederiva F, Rothenberg SS, Hall N, et al. Congenital lung malformations. Nat Rev Dis Primers. 2023;2:9:60. https://doi.org/10.1038/s41572-023-00470-1.
11. Dehner LP, Schultz KAP, Hill DA. Congenital Pulmonary Airway Malformations with a Reconsideration and Current Perspective on the Stocker Classification. Pediatr Dev Pathol. 2023;26:241-9. https://doi.org/10.1177/10935266221146823
12. Le M, Harms P, Peldschus K, Junge CM, Tomuschat C, Reinshagen K. A Series of 40 Congenital Lung Malformation Cases and the Informative Value of CPAM Lesion Ratios. Pediatr Rep. 2025;9;17:5. https://doi.org/10.3390/pediatric17010005

13. López-Díaz M, Cano Novillo I, Morante-Valverde R, et al. Thoracoscopic Lobectomy for Congenital Lung Malformation in Children: Evolving Management Strategies and Their Impact in Outcomes. J Pediatr Surg. 2025;60:161992. https://doi.org/10.1016/j.jpedsurg.2024.161992.
14. Farolfi A, Ghezzi M, Calcaterra V et al. Congenital Lung Malformations: Clinical and Functional Respiratory Outcomes after Surgery. Children (Basel). 2022;9:1881. https://doi.org/10.3390/children9121881.
15. Pardo L, Viveros J, Carrillo J, et al. Manifestaciones radiológicas De Malformaciones Pulmonares congénitas. Experiencia De Tres Hospitales En Bogotá. Rev. colomb. radiol. 2019;30:5117-25. https://doi.org/10.53903/01212095.67
16. Núñez-Paucar H, Atamari-Anahui N, Valera-Moreno C. Congenital pulmonary malformations in children in a pediatric hospital in Peru, 2010-2020. Bol Med Hosp Infant Mex. 2023;80:235-41. https://doi.org/10.24875/BMHIM.23000055.