



Key words (MeSH)

Congenital abnormalities
Multidetector computed
tomography
Lung diseases
Child

Palabras clave (DeCS)

Anomalías congénitas
Tomografía
computarizada
multidetector
Enfermedades
pulmonares
Niño

RADIOLOGICAL MANIFESTATIONS OF CONGENITAL LUNG MALFORMATIONS. EXPERIENCE OF THREE HOSPITALS IN BOGOTÁ

Manifestaciones radiológicas de malformaciones pulmonares congénitas. Experiencia de tres hospitales en Bogotá

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Summary

Objective: To describe the radiological characteristics of congenital pulmonary and airway malformations which are frequently found in pediatric patients, from three hospitals in Bogotá between the years 2010 - 2016. **Materials and methods:** Retrospective, observational and descriptive study with a sample of 27 patients, with an average age of 5 months, who met inclusion criteria: patients between 0 months and 17 years of age, with a confirmed diagnosis of congenital malformation of the lung, who underwent surgery for lung or airway lesion and whose histopathological study was compatible with congenital malformation of the lung. **Results:** The prevalence of congenital malformations is higher in females. 80% of cases had prenatal diagnosis, with cystic adenomatoid malformation being the most common and the main radiological feature being the cyst. **Conclusion:** Computed tomography allows detailed studies of these malformations, achieving greater accuracy compared to conventional techniques such as chest radiography and ultrasound.

Resumen

Objetivo: Describir las características radiológicas de las malformaciones congénitas pulmonares y de la vía aérea que se encuentran frecuentemente en pacientes pediátricos, de acuerdo con la experiencia de tres hospitales de Bogotá, entre 2010 y 2016. **Materiales y métodos:** Estudio retrospectivo, observacional y descriptivo con muestra de 27 pacientes de 5 meses de edad promedio, que cumplieron criterios de inclusión: pacientes entre los 0 meses y 17 años de edad, pacientes con diagnóstico

confirmado de malformación congénita del pulmón, pacientes intervenidos quirúrgicamente por lesión pulmonar o de la vía aérea y cuyo estudio histopatológico fue compatible con malformación congénita del pulmón. **Resultados:** La prevalencia de las malformaciones congénitas es mayor en el sexo femenino, 80 % de los casos contaron con diagnóstico prenatal, la malformación quística adenomatoidea es la más frecuente y la principal característica radiológica es el quiste. **Conclusión:** La tomografía computarizada permite estudios detallados de estas malformaciones, con una mayor precisión en comparación con las técnicas convencionales como radiografía de tórax y ultrasonografía.

1. Introduction

Pulmonary and airway congenital malformations (PACM) form a group of entities originating from bronchopulmonary growth defects during the different evolutionary stages of the lung. They have variable biological behaviour and their prognosis depends on the embryological moment in which the failure and secondary functional alteration originated (1).

The annual incidence of congenital pulmonary malformations is estimated between 32 and 42/100,000 inhabitants (2). The time of diagnosis of PACM is variable: it may be prenatal, postnatal, infancy and, less frequently, in adolescents and young adults. The spectrum of clinical manifestations includes asymptomatic patients, cases similar to mild respiratory infections and severe acute respiratory distress (2,3).

The role of diagnostic images in patients with suspected PACM varies according to the time of diagnosis and associated clinical manifestations. In the prenatal period the approach is performed with ultrasound and can be complemented with magnetic resonance, and in the postnatal period is characterized by chest radiography and computed tomography (CT). PACMs are manifested in imaging studies as parenchymal lesions (opaque or lucid), of variable size and location, according to the specific entity and the time of the study.

2. Material and methods

Series of cases of patients with congenital pulmonary malformation, diagnosed and operated in three institutions of Bogotá, during 4 years. A total of 27 patients were included: 21 with prenatal diagnosis and 6 with postnatal diagnosis. The images were analyzed retrospectively by radiologists with experience in chest radiology and pathologists with expertise in pulmonary diagnosis.

3. Results

The study contains 27 cases: 16 women and 11 men, between 0 days and 17 years old; 21 of them had prenatal diagnosis and in the other 6 the diagnosis was made during the study of patients with repeated respiratory infections or respiratory distress. All cases were evaluated with CT: 17 cystic adenomatoid malformations, 6 pulmonary sequestrations, 2 bronchogenic cysts, 1 scimitar syndrome and 1 pleuropulmonary blastoma.

The main manifestations in imaging studies included: cystic lesion (16), mass (14), consolidation (11) and frosted glass areas (9). Other less frequent findings were: atelectasis, pleural effusion, emphysema and cavitation (Table 1); 21 patients were operated during the first month after imaging diagnosis.

Associated malformations such as diaphragmatic hernias and anomalous venous drains were found. In the reports of pathological anatomy of the specimens, 100 % was consistent with the radiological diagnosis.

Table 1. Findings in thoracic tomography of PACM

Findings	N.º de cases*	Proportion (%)*
Cyst	16	62
Mass	14	54
Consolidation	11	42
Ground glass	9	35
Atelectasia	7	27
Cavitation	2	8
Pleural effusion	2	8
Pneumothorax	2	8
Mosaic attenuation	2	8
Emphysema	2	8
Pulmonary nodule	1	4
Opacity	1	4
Insufflation	1	4

*Several pathologies have associated patterns. Source: Own elaboration.

3.1. Pulmonary cystic adenomatoid malformation

Seventeen cases of patients diagnosed with pulmonary cystic adenomatoid malformation (PCAM) were documented. In 14 of them the diagnosis was suggested by prenatal ultrasound, confirmed by images in the neonatal period and resected before one month of age. In three cases the diagnosis was made by repeated pneumonias before 6 months of life. The types of PCAM confirmed by histology were: PCAM 1 (7 cases), PCAM 2 (8 cases) and PCAM 3 (2 cases).

Lesions were located in the lower left lobe (6 cases), lower right lobe (5 cases), upper left lobe (3 cases), upper right lobe (2 cases) and middle lobe (1 case) (Table 2). All patients underwent thoracotomy and resection of the lesion. Histological examination was consistent with the suggested imaging diagnosis in all cases.

Table 2. Location of PCAM

Location	PCAM 1		PCAM 2		PCAM 3	
	n = 7	%	n = 8	%	n = 2	%
Left lower lobe	-	-	3	38	1	50
Left superior lobe	3	43	-	-	-	-
Right superior lobe	1	14	2	25	-	-
Right middle lobe	1	14	-	-	-	-
Right lower lobe	2	29	3	38	1	50

Source: Own elaboration.

3.2. Pulmonary Sequestration

All the pulmonary sequestrations in the series were extralobar, six cases (3 women and 3 men) had prenatal diagnosis, with resection of the lesion before the month of life, asymptomatic. The most frequent location was in the left lower lobe (3 cases), in the left upper lobe (1 case), in the right middle lobe (1 case) and in the right lower lobe (1 case). Two cases were associated with cystic adenomatoid malformation type 2.

The anomalous arterial supply arose from the thoracic aorta in four cases and from the abdominal aorta in two cases. Venous drainage was only found in two cases: one to the hemiazygos vein and another to a systemic vessel tributary of the inferior vena cava; in three cases the CT scan could not show venous drainage. Other pathologies were found: two cases of bronchogenic cysts, a pleuropulmonary blastoma, and a scimitar syndrome.

4. Discussion

In the studied group there was predominance of female sex 1:0,68. The mean age of diagnosis reported in the literature is less than one year in most PACMs; the age at diagnosis in these patients had a median of 5 months and was as early by prenatal documentation and as late as a case diagnosed at 17 years of age. There were no deaths and 26 cases required surgical treatment. Despite being rare anomalies, it is important to remember that diagnostic suspicion is established on the basis of symptomatology and simple chest radiography in patients ranging from asymptomatic to severely ill.

PACMs include a broad spectrum of abnormalities in the development of the lungs, airway, or pulmonary vascularization. Most cases are detected during the neonatal period or childhood and symptomatology can simulate infectious entities. Diagnostic imaging plays a key role in the detection, classification and follow-up of pulmonary congenital malformations. Conventional radiography is the method indicated in the initial assessment of patients with suspected pulmonary diseases of a congenital nature and in some entities such as diaphragmatic hernia. CT with the possibility of multi-planar and volumetric reconstructions offers additional indispensable information in a significant percentage of patients, due to the possibility of accurately assessing the airway and pulmonary vascularization (3).

This series of cases presents the limitations of observational investigations that analyze data previously recorded in clinical histories. However, this design is very useful in pathologies that are difficult to diagnose, either because of the need for methods that are not within the reach of most patients or because diagnosis is very often accidental due to the lack of clinically important symptoms for patients. Magnetic resonance imaging (MRI) was not requested because the chest CT scan confirmed the diagnosis.

The most frequent pathologies and characteristic radiological findings are summarized below.

4.1. Pulmonary cystic adenomatoid malformation

Pulmonary cystic adenomatoid malformation (PCAM) is a congenital lung mass of unknown cause, resulting from a disordered hamartomatous proliferation and adenomatoid of primary bronchioles that are in communication with the bronchial tree, and receives vascularization of the pulmonary circulation (3,4). According to the literature, the symptoms may be cough, dyspnea, hemoptysis, respiratory distress or even patients may remain asymptomatic; typical manifestations are respiratory distress in the newborn or repeated respiratory infections in older children. In the current series the majority of patients were asymptomatic and only in three cases there were recurrent respiratory infections.

Ch'in and Tang initially described it as a distinct clinical entity (5). In 1977 Stocker and collaborators (6) described these lesions and based on size, shape, macroscopic and histological appearance, made the following classification: type I: 2-10 cm macrocystic; type II: multiple small uniform cysts of 0.5-2 cm; type III: solid-looking lesion with microscopic cysts.

In 2002 Stocker (7) converted I, II, III to 1, 2 and 3, and expanded the classification to include type 0 lesions, previously described as acinar pulmonary dysplasia, which are incompatible with life, therefore, imaging studies are not generally performed (2), and lesions type 4, a kind of peripheral lung cyst that manifests itself with pneumothorax (8) (Table 3).

4.1.1. Type 1 PCAM

It is probably the most common cystic lung lesion, accounting for approximately 60-65 % of all PCAM lesions. It is usually symptomatic in the newborn and manifests as respiratory distress minutes to hours after birth. Smaller lesions may not be evident at birth, but are later discovered if infected. At the end of the first trimester, PCAM 1 can be identified by ultrasonography as a large cystic lesion, which can compress the adjacent lung, move the mediastinum towards the contralateral side and flatten the ipsilateral diaphragm (1,9). Surgical removal of the affected lobe (one lobe in 95 % of cases) allows the other lobes to expand normally and in the absence of other abnormalities the child survives without apparent consequences (10). Seven cases were presented in this series, of which only three had respiratory symptoms and recurrent infections after 3 months of life, all had surgical management and confirmation by histology 100 % consistent with the CT (Figure 1).

Table 3. Stocker Classification

PCAM	Origin	Incidence	Macroscopic characteristics	Microscopic features	Forecast
Type 1	Bronchium	60-70 %	0-2 cm cysts	Larger cysts are usually accompanied by smaller ones. They are covered with pseudostratified columnar epithelium, mucous cells.	Neonates and infants Resectable Good prognosis
Type 2	Bronchus/ Bronchiole	10-15 %	0.5-2 cm small multiple cysts	Small, relatively uniform, bronchiolus-like cysts separated by normal alveoli. The cysts are lined with cuboid to columnar epithelium and have a thin fibromuscular wall.	Neonates Other associated malformations Poor prognosis
Type 3	Bronchiole	5 %	Solid appearance	It resembles the late fetal lung. Mass without obvious cyst formation. Microscopic adenomatoid cysts.	Poor prognosis.

4.1.2. Type 2 PCAM

Approximately 20-40 % of MAQP 2 lesions come from the terminal bronchioles and have two characteristics: cysts in the range of 0.5 to 2.0 cm and their association (50 %) with other abnormalities, such as bilateral renal agenesis, cardiovascular malformations, diaphragmatic hernia and extralobar pulmonary sequestration, which can modify the prognosis of patients (6,11). Eight cases are presented, all with prenatal diagnosis and surgical management before the month of life. Two of the type 2 PCAM cases were associated with extralobar pulmonary sequestration and diaphragmatic hernia (Figure 2).

4.1.3. Type 3 PCAM

It is less frequent and accounts for 10 % of the AQAMs. It is observed as a solid mass accompanied by cysts < 2 mm that can be identified by histology (12). They have an acinar origin and are often very large and cover an entire lobe or several lobes. Two cases with asymptomatic prenatal diagnosis were presented in this series (Figure 3).

In this series, 17 cases with PCAM were studied, the majority of the population was less than 6 months old, male to female ratio 0.7:1. As described in the literature, there is no sex predilection and most had prenatal ultrasound diagnosis. Type 2 PCAM was shown to be the most frequent histologic type, unlike most published series that show type 1 PCAM as the most frequent presentation. The main radiological feature was the variability in cyst size in 13 cases, with predominance in lower lobes. Surgical resection of the lung lesion is the treatment of choice, to avoid recurrent infections and malignant transformation. All cases had good prognosis after surgical correction. Differential diagnoses of cystic chest lesions in a neonate include bronchogenic cyst, congenital lobar emphysema, pulmonary sequestration, congenital diaphragmatic hernia and mediastinal masses, such as neuroblastoma.

4.2. Pulmonary sequestration

Pulmonary sequestration (PS) is a rare disease, of unknown etiology, accounting for 0.1-6 % of all structural lung diseases and

developmental malformations. During the fourth week of gestation a lung-like structure develops without communication with the main bronchial tree, envelops itself with its visceral pleura and derives its arterial supply from the systemic circulation (13).

In 1946, Pryce (14) used the term “pulmonary sequestration” and classified the disease as intralobar and extralobar. An intralobar sequestration (ILS) is the most common form; in this the lesion remains within the visceral pleura of his lobe, while the extralobar sequestration (ELS) type corresponds to a true accessory pulmonary tissue, with a separate pleural sheath and aberrant venous drainage; depending on the segment from which the accessory pulmonary bud branches out of the primitive anterior bowel, the sequestration is subclassified as intrathoracic (supradiaphragmatic) or abdominal (subdiaphragmatic) type (15).

Yong et al (16) retrospectively evaluated 2,625 cases in China, as follows: male-female ratio of 1.58 (1,873 cases of ILS 83.95 % and 358 cases of ELS 16.05 %), bilateral cases were rare. The majority presented symptoms such as cough, sputum, fever, hemoptysis and chest pain, 13 % were found asymptomatic.

Chest CT showed mass type lesions in 49 %: cystic lesions in 29 %, cavitated lesions in 12 % and other lung lesions in 8 %. Sequestration was mainly located in the lower lobe. Arterial supply was mostly provided by branches of the thoracic aorta (77 %) and abdominal aorta (18 %). Venous drainage of pulmonary sequestration was performed on pulmonary veins in 91 %.

In the series of this article all seizures were extralobar - contrary to what is reported in the literature, which shows intralobar pulmonary seizure as the common pathology - located in the lower left lobe in half of the cases, all asymptomatic. The initial radiological presentation in 5 cases was intrapulmonary mass, they received irrigation from the thoracic aorta and presented venous drainage to the hemiaeous vein. One case presented right pneumothorax with closed thoracotomy requirement.

Associated malformations were found in 2 cases: a right diaphragmatic hernia and a type 2 PCAM. Surgical treatment should be considered for most patients, especially symptomatic patients (16,17) (Figure 4).

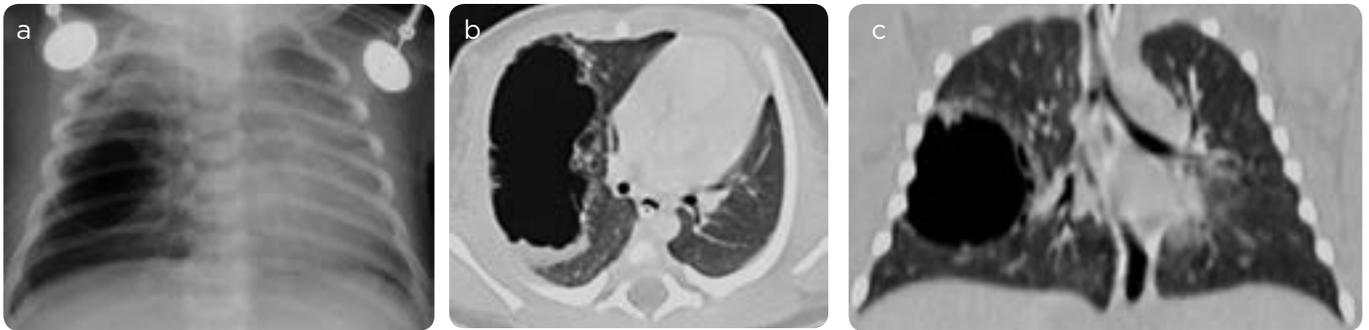


Figure 1. Type 1 cystic adenomatoid malformation. 5-day-old girl. a) Chest X-ray. Radiolucent image of round morphology and thin wall in the right lung. b and c) CT scan of chest, axial and coronal reconstruction. Cystic lesion in the middle lobe, lobed contours and thick wall.

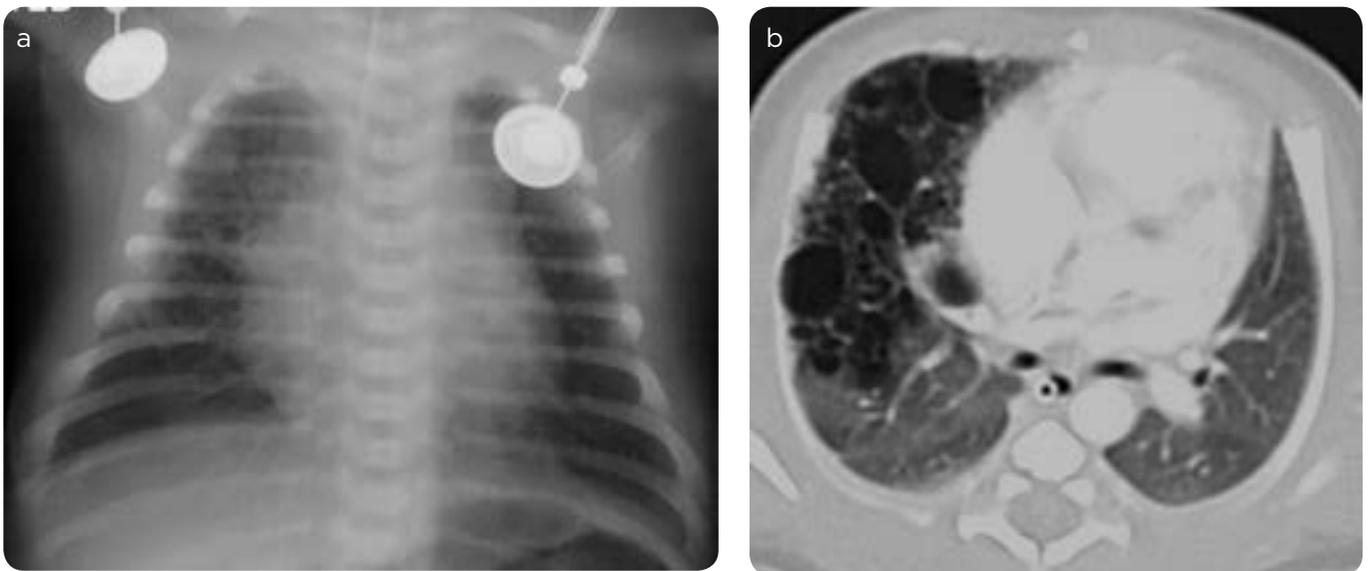


Figure 2. Adenomatoid cystic malformation type 2. Child 4 days old. a) Chest X-ray. Thin-walled radiolucent lesions in the right lung. b) Chest CT, axial. Upper and middle lobe mass consisting of multiple cystic-looking, thin-walled lesions.

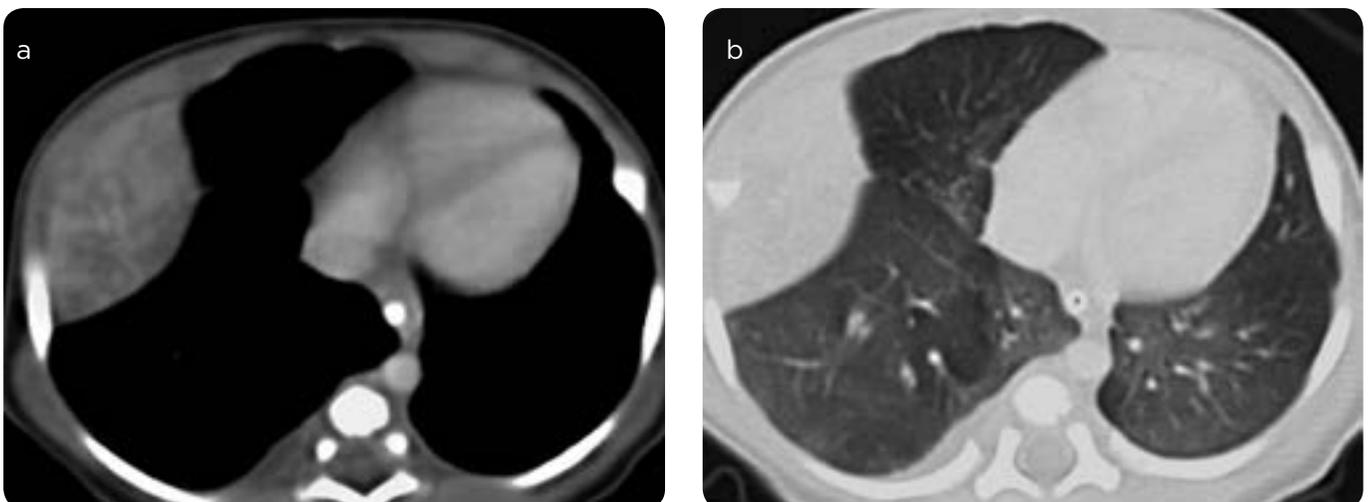


Figure 3. a and b). Type 3 cystic adenomatoid malformation. 2-year-old girl. CT scan of chest, axial, mediastinal window and pulmonary window. Mass in the lower right lobe, broad pleural base, solid, with pointed cystic areas.

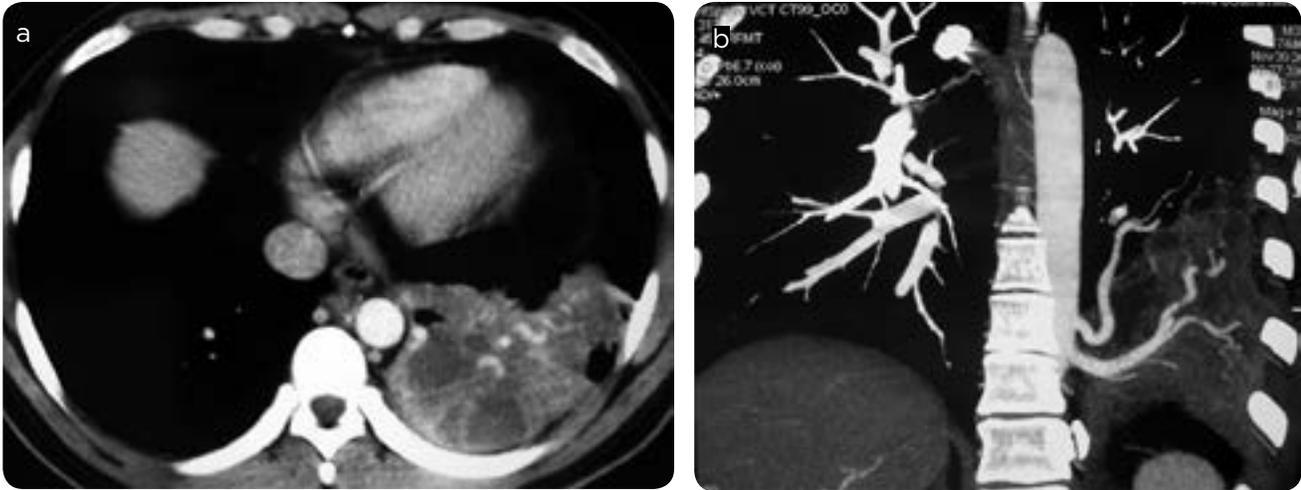


Figure 4. Pulmonary Sequestration. Child 4 years old. a and b) Chest CT, axial, in mediastinal window and coronal reconstruction MIP. Mass in left lower lobe of heterogeneous density with systemic irrigation.

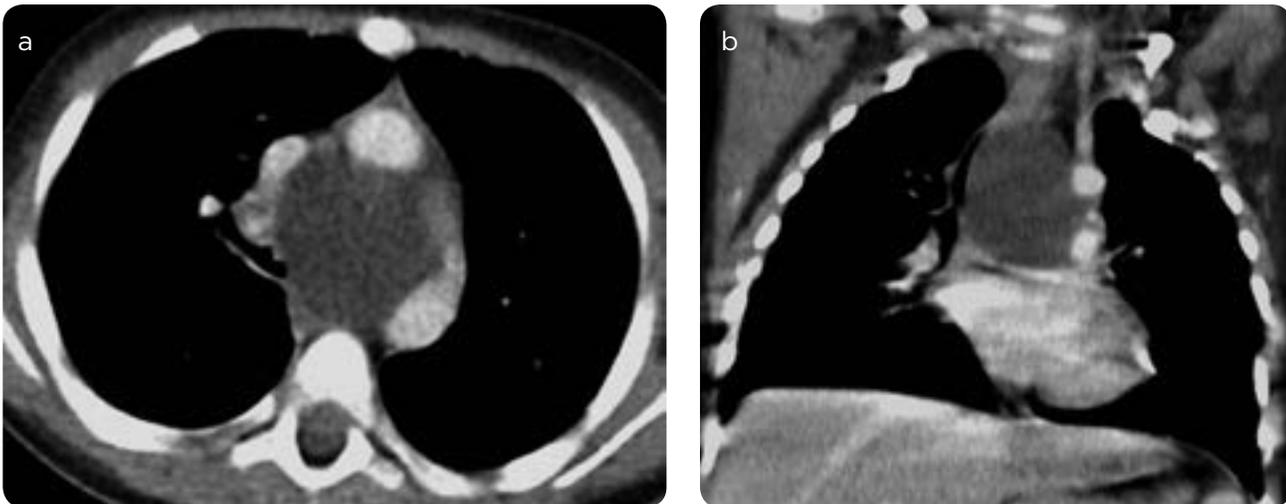


Figure 5. Bronchogenic cyst. 1-year-old girl. a and b) Chest CT, axial and coronal reconstruction in mediastinal window. Subcarinal mediastinal mass, cystic, thin-walled, without enhancement.

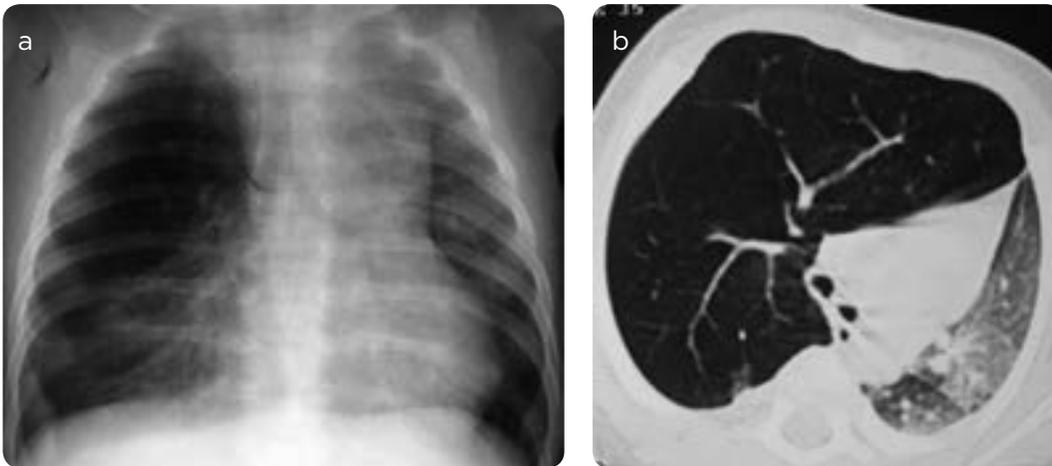


Figure 6. Congenital lobar emphysema. 7-month-old girl. a) Chest x-ray. Radiolucency and hypovascularity in the right pulmonary field. b) CT scan of chest, axial, pulmonary window. Hyperinflation of the right upper lobe with contralateral displacement of the mediastinal structures and passive atelectasis of the left pulmonary parenchyma.

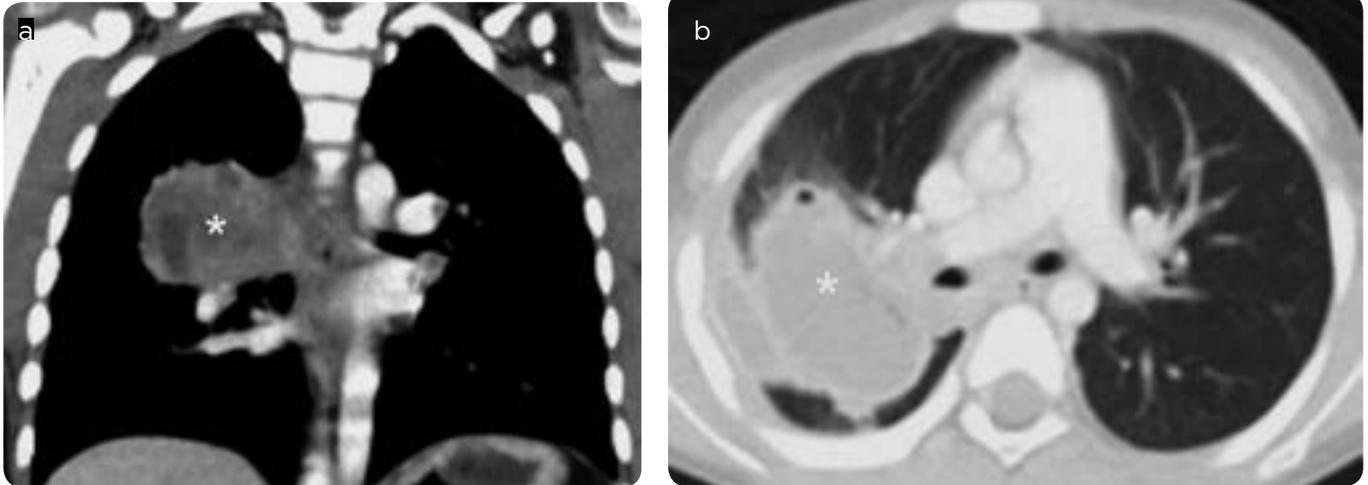


Figure 7. Pleuropulmonary blastoma. 3-year-old girl. a and b) CT scan of chest, coronal, mediastinal and pulmonary window. Solid-looking mass, lobed contours and heterogeneous density in the posterior segment of the right upper lobe.



Figure 8. Scimitar syndrome. 6-year-old girl. Chest CT scan, coronal MIP reconstruction. Tubular structure in the right pulmonary field corresponding to anomalous venous drainage of the right lower lobe.

4.3. Bronchogenic cyst

Bronchogenic cysts (BC) are congenital lesions. They are thought to originate from the primitive ventral anterior bowel and may be mediastinal or intrapulmonary. Approximately two-thirds are within the mediastinum and one-third are intraparenchymal (18). The mediastinal location can be paratracheal (generally, on the right side), carinal, the most frequent, or hilar. They must be differentiated from cystic teratomas, thymic cysts or ectopic thyroid glands (19). Most are asymptomatic, but occasionally cause symptoms secondary to compression of adjacent structures. These symptoms include chest pain, cough, dyspnea, dysphagia, fever, and purulent sputum (20,21).

Rogers and Osmer (22) described the radiographic findings of a bronchogenic cyst as a clearly defined, solitary, non-calcified, round or oval density divided into three categories: a cyst with a water density, an air-filled cyst, or a cyst containing a hydro-air level, when infected cysts

may show a wall enhancement. CT has the ability to locate an intrathoracic cyst, and define its extent and relationship to nearby structures.

Two symptomatic cases are shown in this article: the first, a 1-year-old girl with bronchogenic cyst in the posterior and superior mediastinum (Figure 5), and the second, a 1-year-old boy with bronchogenic cyst in the middle mediastinum next to the aortic arch, both with surgical management and diagnostic confirmation with an anatomopathological study.

4.4. Congenital lobar emphysema or pulmonary hyperinflation

Congenital lobar emphysema (CLE) is a rare disorder of lung development, but is due to partial obstruction of the lobar bronchus, resulting in hyperinflation of the affected lobe with irreversible destruction of the alveolar septa (23,24). The incidence of CLE is possibly underestimated, varies between 1:20,000 and 1:30,000 births, the male to female ratio is 3:1 (25,26). The left upper lobe and the middle lobe are the usual sites, the lower lobes are rarely affected. Respiratory impairment usually occurs in the first few days of life, and 90 % of cases occur before the age of 6 months (27). CT is performed to confirm the diagnosis, evaluate mediastinal vascular structures and rule out other abnormalities; it shows a radiolucent lung with scarce vessels, mass effect with mediastinal displacement, frequent in the upper left lobe (28).

The only case in this series shows the same pattern as described in the literature, with symptomatic right upper lobe involvement after 6 months of age. Surgery was performed and the anatomopathological study of the piece confirmed the initial diagnosis (Figure 6).

4.5. Pleuropulmonary blastoma

Pleuropulmonary blastoma (PPB) is a rare primary malignant pulmonary tumor that occurs exclusively in pediatric age, especially in children younger than 6 years of age. It belongs to a unique category of tumors observed exclusively in childhood: dysembryogenic neoplasms, analogous to Wilms' tumor, neuroblastoma and hepatoblastoma (29,30). It was first described in 1952 by Barnard (31). It is usually located on the periphery of the lung, but may be extrapulmonary, and affects the parietal pleura, mediastinum, great thoracic vessels, regional lymph nodes, and diaphragm.

PPB is of mesodermal origin, classified by Dehner and collaborators (32) in 1994 in three histological types: cystic (type I) the most frequent, affects children under 10 months; mixed (type II) with solid-cystic component, and solid (type III), of poor prognosis, affecting people over 4 years of age. The higher the histological type, the higher the mortality, so that the clinical course will depend fundamentally on the anatomopathological result (33).

In the case series of Priest and collaborators (30) evaluated 50 cases, respiratory distress with or without fever was the most frequent clinical symptom. The formation of cysts in the affected lung was identified radiographically in 19 children (38 %) before the definitive pathological diagnosis. The ages of manifestation of PPB cases were: 7 type I cases (10 months), 24 type II cases (34 months) and 19 type III cases (44 months). PPB type I is the least common of the three tumor types and occurs in very young children, including newborns.

In this series metastases occurred in 13 cases with PPB type II or type III, mainly in the brain and bone. Five-year survival was 83 % for type I and 42 % for types II and III.

Images show a large mass in the thorax with a solid-cystic aspect, associated with pleural effusion, contralateral mediastinal displacement without invasion of the thoracic wall (34). There are no conclusive studies for the treatment of this disease, surgery, chemotherapy and radiotherapy may be offered when resection is not complete, but none prevents local recurrence or metastasis. The only case in the series of this article with this diagnosis is a type III PPB (Figure 7).

4.6. Scimitar syndrome

Scimitar syndrome is characterized by hypoplasia of the right lung, dextroposition of the heart, and abnormal drainage of the pulmonary vein in the inferior vena cava, which produces a curved vascular shadow along the right edge of the heart (35). The partial anomalous venous connection of the pulmonary vein drains systemic veins, inferior vena cava and, less commonly, the portal vein, hepatic veins, right atrium (36). Three forms of scimitar syndrome have been described. In the infantile form there is a large shunt between the abnormal artery that irrigates the lower lobe of the right lung and the subdiaphragmatic aorta; this is sometimes called hijacking. In the adult form there is a small short circuit between the right pulmonary veins and the inferior vena cava. The third form is associated with cardiac and extracardiac malformations.

Patients may remain asymptomatic, or be detected incidentally or develop symptoms of congestive heart failure or recurrent right basal pneumonia. Chest x-rays typically show a hypoplastic right lung, which may be associated with ipsilateral mediastinal displacement and hyperinflation of the contralateral lung, the pulmonary vein that descends along the right cardiac rim is characteristic and corresponds to the sign of the scimitar (37). Right pulmonary vein anomaly can be identified as a curvilinear opacity in the right lower thorax, which can be delineated on CT or MR with angiography, along with any associated systemic anomaly (38). The series of this article documents a case of scimitar syndrome that did not require surgical treatment (Figure 8).

5. Conclusions

CT plays a key role in the assessment of a significant percentage of patients with CPM and in some entities, such as pulmonary sequestration,

its performance may be essential. The high doses of ionizing radiation inherent in CT should be controlled with low-dose protocols with automatic tube modulation in the pediatric patient and new techniques, such as reconstruction, that allow diagnostic quality studies with significantly lower doses of radiation. The use of IV contrast medium is mandatory due to the possibility of pulmonary sequestration or hybrid malformations with PACM histology associated with anomalous systemic vessels.

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Received for assessment: October 16, 2018

Accepted for publication: April 12, 2019