

# Management and follow-up of congenital lung malformations

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## ABSTRACT

**Objective:** Congenital lung malformations (CLM)—including congenital pulmonary airway malformation (CPAM), bronchopulmonary sequestration (PS), congenital lobar overinflation (CLO), bronchogenic cyst (BC), and isolated congenital bronchial atresia (BA)—have shown increased prenatal diagnosis rates with the more effective use of antenatal imaging methods such as ultrasound (US) and magnetic resonance imaging (MRI). However, the optimal management of these lesions remains unclear. We aimed to investigate the diagnostic processes and management approaches in patients with CLM diagnosed at our clinic.

**Material and Methods:** This retrospective, cross-sectional study included 46 patients aged 0–18 years who were diagnosed with CLM between April 2018 and March 2025. Data on the time and method of diagnosis, presenting complaints, whether surgery was performed, and imaging modalities were recorded.

**Results:** The median age of the patients was 60 months (IQR 62). In the prenatal diagnosis group, the median age at first presentation was 38 months (min–max=1–156) for asymptomatic patients and 6 months (min–max=0.2–132) for symptomatic patients. Sixty-three percent of cases were diagnosed prenatally, and 37% were diagnosed postnatally. Sixty-nine percent (20/29) of prenatally diagnosed cases were asymptomatic at birth. Most radiological lesions were unilobar and unilateral and did not show mediastinal shift. The most commonly affected lobe was the right lower lobe. CPAM (43.5%) was the most common diagnosis, followed by PS (17.8%). Advanced imaging, in addition to ultrasound and chest X-ray, was performed in 20% of the CPAM group and 37.5% of the PS group.

**Conclusion:** The majority of our CLM cases were diagnosed prenatally. In prenatally diagnosed cases that are asymptomatic postnatally, the types and timing of diagnostic tests vary on a case-by-case basis, and the timing of surgical treatment remains uncertain. We believe that further prospective studies and stronger scientific evidence are needed in this field and that management decisions should be based on an individualized benefit–risk assessment.

**Keywords:** Computed tomography, congenital lung malformations, diagnosis, surgical treatment.

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## INTRODUCTION

Congenital lung malformations (CLM) include congenital pulmonary airway malformation (CPAM), bronchopulmonary sequestration (PS), congenital lobar overinflation (CLO), bronchogenic cyst (BC), and bronchial atresia (BA). These conditions develop from malformations of the lung parenchyma, airways, and vascular structures.<sup>[1]</sup>

Advancements in diagnostic methodologies, most notably prenatal ultrasound and fetal magnetic resonance imaging (MRI), have enabled the detection of the majority of congenital lung malformations prior to birth. Fetal MRI is a complementary imaging modality that provides clearer anatomical details of the lesion and aids in differential diagnosis from other thoracic anomalies. The management of CLM during the prenatal period is contingent upon the size of the lesion, its growth rate, and its effects on the fetus. Postnatal management depends on the symptomatology of the lesion and its dimensions. This is of great importance for planning postnatal management and preventing potential complications. Symptoms vary; therefore, lesions may be detected at birth with respiratory distress and cyanosis or incidentally in asymptomatic adults.<sup>[1]</sup> It is of paramount importance to diagnose and treat patients with severe respiratory distress in a timely manner, as this condition is often accompanied by high morbidity and mortality rates. Early diagnosis of CLM is imperative for optimal prenatal counselling and early perinatal and postnatal management.<sup>[2,3]</sup>

The enhanced efficacy of antenatal imaging techniques, such as ultrasonography (US) and, more recently, magnetic resonance imaging (MRI), has resulted in an increase in the prenatal diagnosis rate. Nevertheless, the optimal approach to the management of these lesions remains to be elucidated, given the heterogeneity of the lesions and the varied clinical presentations.<sup>[1,3]</sup>

The decision-making process can be challenging for clinicians because of the complex structure and diversity of these malformations. In our study, we examined the diagnostic processes and approaches used in patients diagnosed with CLM. The aim was to discuss ideal management strategies in the context of current healthcare conditions in our country.

## MATERIAL AND METHODS

This retrospective, cross-sectional study included 46 patients aged 0–18 years who were diagnosed with CLM and followed up between April 2018 and March 2025 at the Department of Paediatric Pulmonology, Güztepe Prof. Dr. Süleyman Yalçın City Hospital, İstanbul Medeniyet University. Information regarding the participants' time of diagnosis, diagnostic method, presenting complaints, surgical history, imaging studies, and follow-up was obtained from the institutional patient information system and handwritten medical records.

Additionally, the following imaging studies were reviewed using the hospital database and the national online health monitoring system (e-Nabız): chest X-ray, ultrasound (US), chest computed tomography (CT), computed tomography angiography (CTA), and magnetic resonance imaging (MRI).

Patients were categorised into four groups based on the timing of diagnosis and symptom status: prenatal diagnosis with symptoms,

prenatal diagnosis without symptoms, postnatal diagnosis without symptoms, and postnatal diagnosis with symptoms.

This study was conducted in accordance with the principles of the 1964 Helsinki Declaration and its subsequent amendments, which establish ethical standards for research involving human subjects. Ethical approval was obtained from the Clinical Research Ethics Committee of İstanbul Medipol University on 22.05.2025 (decision number: 644).

## Statistical Analysis

Data were analysed using SPSS version 25.0 software. The normality of data distribution was assessed using the Shapiro–Wilk test. Owing to the descriptive nature of the study, only descriptive statistics were performed. Categorical variables were expressed as numbers and percentages. Continuous variables were presented as median (interquartile range (IQR), minimum–maximum) for non-normally distributed data. No inferential statistical analyses were performed.

## RESULTS

The median age of the 46 patients included in the study was 60 months (interquartile range [IQR] 62), and the median age at first presentation to the clinic was 17 months (minimum–maximum [min–max]=0.2–156.0 months). The median follow-up period at our clinic was 50 months (IQR 47.8). At the time of initial presentation, data according to diagnostic groups were as follows: for prenatally diagnosed asymptomatic patients, the median age was 38 months (range 1–156 months); for prenatally diagnosed symptomatic patients, the median age was 6 months (range 0.2–132 months); for postnatally diagnosed asymptomatic patients, the median age was 14 months (min–max=8–148 months); and for postnatally diagnosed symptomatic patients, the median age was 26 months (min–max=1–132 months).

The demographic profile of the cohort showed a predominance of males, accounting for 56.5% (n=26) of patients. The majority were born at term, and no parental consanguinity was documented in most cases. Parental consanguinity was present in 22.7% (n=10) of patients, while 9.3% (n=4) were born preterm. Overall, 63% of cases were diagnosed prenatally and 37% postnatally. Among prenatally diagnosed cases, 69% (20/29) were asymptomatic at birth, whereas 31% (9/29) were symptomatic. Most radiological lesions were unilobar and unilateral. Congenital heart malformations were present in 21.7% of cases. The most frequently affected lobe was the right lower lobe. Based on symptom status and timing of diagnosis, the most common groups were asymptomatic before birth (43.5%) and symptomatic after birth (30.4%). The most prevalent lesion type was CPAM (43.5%), with type 2 being the most common subtype (Table 1).

Operative age according to diagnostic group was as follows: asymptomatic patients with prenatal diagnosis had a median operative age of 18 months (IQR 51); symptomatic patients with prenatal diagnosis had a median operative age of 0.6 months (IQR 0.8); asymptomatic patients with postnatal diagnosis had a median operative age of 82 months (IQR 64); and symptomatic patients with postnatal diagnosis had a median operative age of 21 months (IQR 81).

**Table 1: Descriptive statistical information related to measurement values (n=46)**

	n	%
Classification by timing and symptom status		
i. Prenatal	29	63.0
Asymptomatic	20	69.0
Symptomatic	9	31.0
ii. Postnatal	17	37.0
Asymptomatic	3	17.6
Symptomatic	14	82.4
Type of CLM		
Bronchogenic cyst	3	6.5
CPAM	20	43.5
Type 3	1	4.3
Type 2	8	34.8
Type 1	6	26.1
N/A	8	34.8
PS	8	17.4
Intrapulmonary	3	30.0
Extrapulmonary	2	20.0
N/A	3	30.0
CLO	4	8.7
Hybrid lesion	2	4.3
CPAM type 1 + PS	1	50
CPAM type 1 + Bronchogenic cyst	1	50
Radiological features		
Anatomical location involved		
Right lower lobe	13	28.3
Right middle lobe	4	8.7
Mediastinum	3	6.5
Left upper lobe	10	21.7
Right upper lobe	4	8.7
Left lower lobe	7	15.2
More than. one lobe	5	10.9
Mediastinal shift	6	13
Extent of affected area		
Single lobe and/or confined to one hemithorax	41	89.1
Multilobar and/or involving both hemithoraces	5	10.9
Coexisting congenital malformations		
I. Gastrointestinal system (TOF)	1	9.1
II. Cardiovascular system (VSD, Dextroposition, ASD, PFO)	10	90.9

CLM: Congenital lung malformation; CPAM: Congenital pulmonary airway malformation; PS: Pulmonary sequestration; CLO: Congenital lobar overinflation; TOF: Tracheoesophageal fistula; VSD: Ventricular septal defect; ASD: Atrial septal defect; PFO: Patent foramen ovale; N/A: Not applicable.

**Table 2: Comparison of radiological preliminary and definitive diagnoses in operated patients (n=28)**

	n	%
Definitive diagnosis		
Bronchogenic cyst	3	10.7
CPAM	14	50
Pulmonary sequestration	7	25
Congenital lobar overinflation	2	7.1
Hybrid lesion	2	7.1
Radiological preliminary diagnosis		
Bronchogenic cyst	3	10.7
CPAM	7	25
Pulmonary sequestration	1	3.6
Air trapping	2	7.1
Cavitary lesion/ consolidation	2	7.1
Solid mass	2	7.1
CPAM	1	3.6
Pulmonary sequestration	4	14.3
Cavitary lesion	2	7.1
Air trapping	2	7.1
Cavitary lesion	1	3.6
Cystic lesion	1	3.6

CPAM: Congenital pulmonary airway malformation.

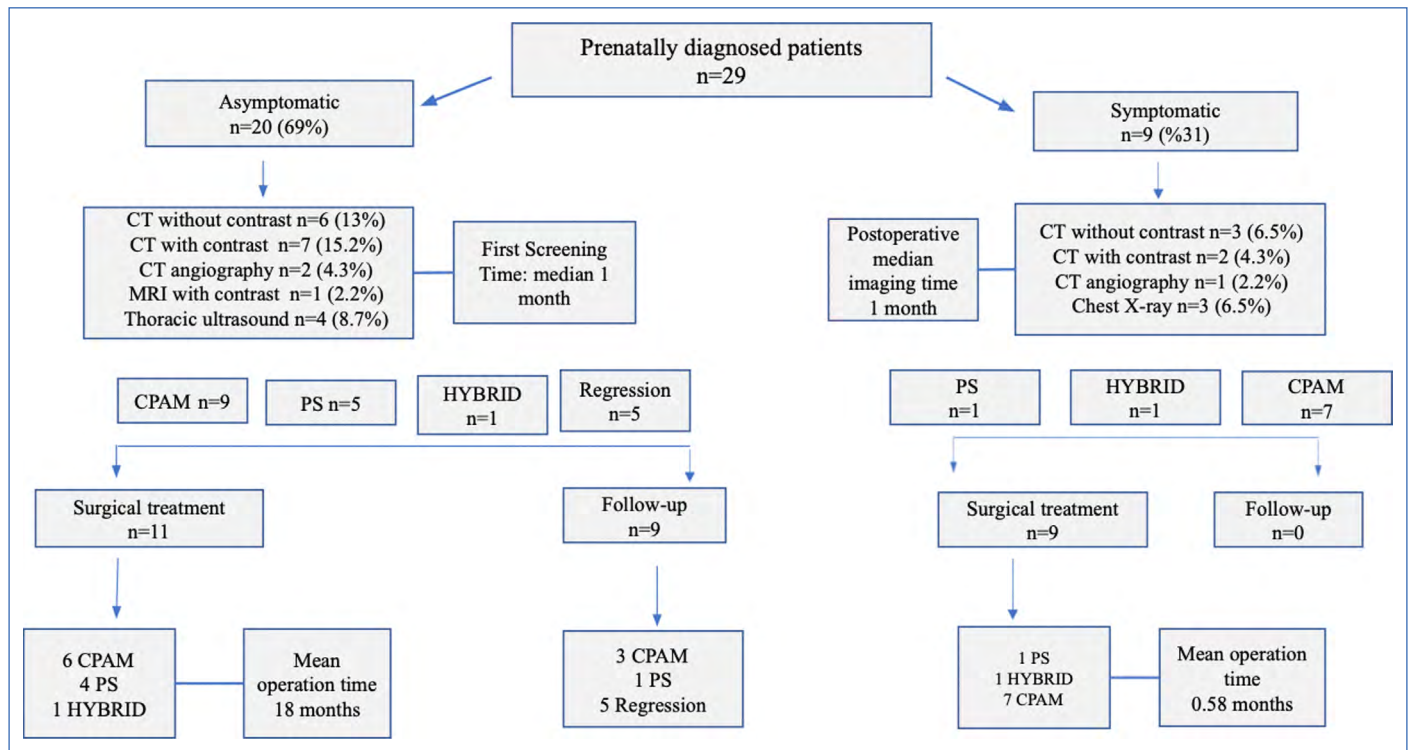
The timing of the first chest X-ray according to diagnostic groups was as follows: prenatally diagnosed asymptomatic patients had a median age of 1.0 month (IQR 29); prenatally diagnosed symptomatic patients had a median age of 1.0 month (IQR 0.5); and postnatally diagnosed asymptomatic patients had a median age of 4.2 months (IQR 3.7).

Among prenatally diagnosed asymptomatic patients, 17.2% (5/29) had a CLM diagnosis that could not be confirmed postnatally, suggesting possible lesion regression (Fig. 1).

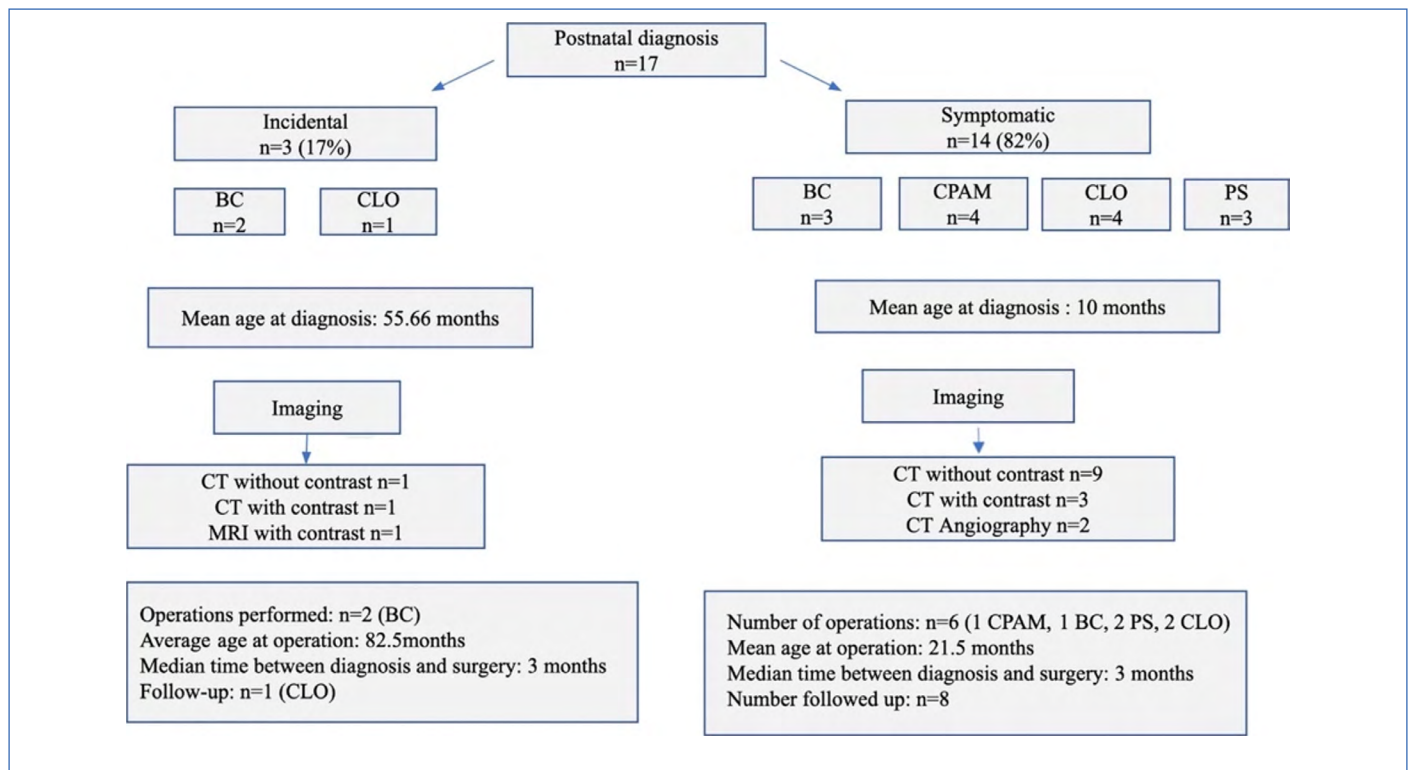
Similarly, 17.6% (3/17) of postnatally diagnosed patients were diagnosed incidentally. These incidental diagnoses included one case of CLO identified on a CT scan performed after trauma, one case detected on contrast-enhanced MRI during evaluation for neurofibromatosis type 1, and one case diagnosed on imaging performed for suspected foreign body aspiration (foreign body aspiration was not confirmed by bronchoscopy in this patient). The mean age at diagnosis in these cases was 55.6 years. Among postnatally diagnosed patients, 82.4% (14/17) were diagnosed based on imaging studies performed for recurrent respiratory symptoms, with a mean age at diagnosis of 10 months (Fig. 2).

### Imaging Findings

Lesions were confined to a single lobe and/or hemithorax in 89.1% of cases, involved multiple lobes and/or bilateral hemithorax in 10.9% of cases, and were associated with mediastinal shift in 13% of cases (Table 1).



**Figure 1:** Follow-up and management of our patients diagnosed with CLM during prenatal screening.



**Figure 2:** Follow-up and management of our patients diagnosed with CLM during postnatally screening.

Comparison of initial radiological diagnoses with final diagnoses showed that CPAM had the highest misdiagnosis rate. Among 14 cases with histopathologically confirmed CPAM after surgery, CPAM was

not considered in 7 cases (50%) during preoperative evaluation, and alternative preliminary diagnoses were made (Table 2). These included pulmonary sequestration, air trapping, cavitory lesion, solid mass, and

consolidation. Among patients who underwent advanced imaging in addition to ultrasound and chest X-ray, 20% of the CPAM group (two MRI and two CT scans) and 37.5% of the PS group (one MRI, one CT scan, and one CT angiography) received advanced imaging.

Surgical treatment was performed in 43.4% (20/46) of all cases, including 37.9% (11/29) of prenatally diagnosed patients and 47% (8/17) of postnatally diagnosed patients. Segmentectomy was performed in 15% (3/20) of cases, mediastinal cyst excision in 10% (2/20), and lobectomy in 75% (15/20), including one case that required additional vascular embolisation. At the time of data collection, eight patients were awaiting surgery as part of the planned surgical management, with follow-up close to their initial presentation.

## DISCUSSION

This article analyses the diagnostic processes of 46 CLM patients who were followed at our clinic for seven years. The majority of patients were diagnosed during the prenatal period (63%), were asymptomatic at birth (69%), and underwent surgery at a median age of 18 months.

The most common diagnostic groups were CPAM and PS, respectively. Regression of the lesion was observed in 10.9% of patients diagnosed IU in the postnatal period. CLMs exhibit diverse clinical characteristics; they may present with severe respiratory symptoms at birth, remain asymptomatic, or be detected incidentally at a later age.

Due to their wide clinical spectrum and albeit low risk of malignant transformation, CLMs require timely diagnosis and appropriate management. Therefore, early diagnosis, prenatal counselling, and early perinatal and postnatal management are essential.<sup>[2,3]</sup> In recent years, an increase in the frequency of prenatal diagnosis has been reported, with up to 4 in 10,000 pregnancies detected during the prenatal period. This increase is primarily attributed to technical advances in fetal ultrasound (US) and magnetic resonance imaging (MRI), as well as their more widespread use.<sup>[4]</sup>

Congenital thoracic malformations (CTMs) are frequently detected during routine antenatal ultrasonography and often encompass a spectrum of anomalies rather than isolated abnormalities. In addition, due to the similar appearance of various congenital lung and non-pulmonary lesions, a definitive antenatal diagnosis is usually not possible. Prenatal diagnosis is generally less reliable than postnatal diagnosis, as it typically relies on the cystic or solid appearance of lesions within the fetal thorax or abnormal lung size, which may result in displacement of the heart.<sup>[5]</sup>

Because our study included patients referred from different maternity clinics, we were unable to evaluate prenatal diagnostic characteristics in detail. Nevertheless, we found that 63% of all CLM cases had received a prenatal diagnosis. Prenatal CLM diagnosis should be confirmed postnatally, and the lesion type must be clearly defined. During fetal ultrasound follow-up, resolution (11–49%), regression (18–42%), or progression (33–44%) of lesions may be observed.<sup>[6]</sup> Cavoretto et al.<sup>[7]</sup> demonstrated that only 34 (44.7%) of 76 cases with prenatal sonographic resolution were confirmed postnatally. The reduction in CLM size appears to be related not only to fetal growth but also to the transition in lung development from

the canalicular to the saccular stage. In addition, in late pregnancy, lesions may become isoechoic with normal lung tissue, leading to the erroneous assumption that they have disappeared.<sup>[6]</sup>

Among prenatally diagnosed patients in our cohort, 17% had no detectable lesions on postnatal evaluation. A prospective study reported spontaneous regression in 17% of 29 cases diagnosed with prenatal CLM, with postpartum confirmation also performed. Prenatal and postnatal diagnoses were consistent in 37% of cases (with partial regression in 25%), whereas incorrect diagnoses, such as congenital diaphragmatic hernia or duplication cyst, were reported in 10% of cases.<sup>[8]</sup> As we were unable to review antenatal imaging, we hypothesised that regression may have occurred in this 17% subgroup, given the absence of detectable lesions and other anomalies. The reported accuracy of prenatal ultrasound in distinguishing lesion types ranges between 60% and 90%.<sup>[9]</sup> Therefore, caution is warranted to avoid unnecessary or inappropriate prenatal and/or postnatal interventions in every infant diagnosed with CLM before birth.

The average age of patients at our paediatric pulmonology centre was 17 months. The youngest patient was younger than 1 month, and the oldest was 156 months. This wide age range was attributable to diagnoses made both prenatally and postnatally. Asymptomatic patients diagnosed prenatally had a mean age of 38 months, whereas symptomatic patients had a mean age of 6 months. These ages were considered relatively late for the initiation of paediatric pulmonology follow-up. The most important contributing factor was referral from other clinics, resulting in delayed presentation. We would like to emphasise the importance of effective inter-clinic communication for timely diagnosis and management.

The group with the greatest uncertainty in management, for which standards are still being developed, consists of asymptomatic patients diagnosed prenatally. Advanced thoracic imaging should be performed to confirm the diagnosis and to evaluate the lesion more accurately in asymptomatic infants; however, the timing depends on individual risk factors. Even if they are asymptomatic postnatally, infants diagnosed prenatally should first undergo a chest X-ray. If a suspicious lesion is detected, chest computed tomography (CT) should be performed.<sup>[10]</sup>

Chest CT is recommended at approximately 6–9 months of age, even in asymptomatic patients, because of the risk of recurrent lung infections and malignant transformation. Chest magnetic resonance imaging (MRI) may also be used in centres with appropriate experience.<sup>[11]</sup> In addition, immediate advanced imaging evaluation is recommended for infants with features suggesting an increased risk of complications, such as large lesions on chest radiography, bilateral or multifocal cysts, or a family history of conditions associated with pleuropulmonary blastoma or pneumothorax. In infants without risk factors associated with poor prognosis, advanced thoracic imaging is recommended at 6–9 months postnatally.<sup>[12]</sup>

All infants in our cohort, whether asymptomatic or symptomatic, underwent chest X-ray evaluation within the first month after birth. Difficulties were encountered in postnatal management because of insufficient information regarding prenatal diagnostic methods. The largest diagnostic group consisted of asymptomatic patients diagnosed prenatally, with the first CT and/or MRI imaging performed

at a mean age of six months. Most patients had previously undergone non-contrast CT imaging. However, contrast-enhanced CT had to be repeated in patients requiring differential diagnosis of other congenital malformations, which occurred in six cases. Contrast-enhanced thoracic CT is recommended to definitively confirm the absence of pulmonary lesions within the first six months, and CT angiography (CTA) is recommended to evaluate associated hybrid lesions.<sup>[13]</sup>

Thoracic CT was the most frequently used imaging modality in our cohort, followed by CT angiography and MRI. Because of the high prevalence of CPAM and PS, advanced imaging was often required for differential diagnosis. The currently accepted view is that CT angiography is necessary to exclude or confirm PS, as the most common hybrid lesions involve the coexistence of CPAM and PS.<sup>[13]</sup>

In the postnatal period, children diagnosed due to respiratory symptoms are treated with early total surgical resection. Reported complications in patients who do not undergo surgery include cyst infection, dyspnoea, pneumothorax, bleeding, feeding difficulties, sudden respiratory failure (affecting approximately 20% of children younger than one year), and malignant transformation (reported in 1–3% of patients). Early surgical resection is also recommended when the lesion involves more than 20% of the hemithorax, in the presence of pneumothorax, bilateral or multifocal cysts, or a family history of pleuropulmonary blastoma. All of our patients diagnosed symptomatically in the postnatal period had recurrent lower respiratory tract infections and no history of malignancy. In most cases, lesions were unilateral and confined to a single lobe. Only one in ten patients had multilobar or bilateral involvement with mediastinal shift, and these cases underwent surgery at a median age of 21 months. Lobectomy was the most commonly applied surgical technique in our series. Following total resection, postoperative survival is generally high regardless of the timing of surgery.<sup>[14]</sup> The optimal timing of surgery in asymptomatic infants remains controversial; however, as anaesthetic and surgical risks decrease after the first few months of life, surgery is generally recommended between six months and two years of age.<sup>[15,16]</sup>

Half of the asymptomatic patients diagnosed prenatally in our cohort underwent surgical treatment, with a median operative age of 18 months. The remaining CLM cases currently under follow-up have not yet been classified as asymptomatic or high-risk lesions and are expected to reach one year of age.

As our study has shown, the most common type of CLM is CPAM, followed by PS. CPAM is characterised by the formation of one or more cysts within the lung parenchyma, typically involving a single lobe. It can be confused with many other malformations or cystic diseases.<sup>[17]</sup> In contrast, pulmonary sequestration can easily be overlooked or confused with other pathologies because it may appear as a solid nodule or mass, consolidation, or cystic lesion on chest X-ray.<sup>[18]</sup>

When we compared the initial radiological diagnoses of our patients with their final diagnoses, CPAM was found to have the highest misdiagnosis rate. Of the 14 cases in which CPAM was confirmed histopathologically after surgery, CPAM was not considered in 50% of preoperative evaluations, and alternative preliminary diagnoses, such as PS, cystic and cavitory lesions, or air trapping, were made. Among the cases in which the final diagnosis was PS, one case had previously been diagnosed as

CPAM and two cases as cavitory lesions. In a prospective study of fetuses with prenatally diagnosed CLM, 5% of lesions regressed spontaneously during the intrauterine period, 8% decreased in size, and 11% were associated with respiratory distress in the neonatal period. In addition, 83% of infants who underwent postnatal CT scanning proceeded to surgery. These infants were followed up until childhood, with a median follow-up period of five years. During follow-up, 5% developed symptoms, such as pneumonia, and underwent surgery between the ages of three and five years. In contrast, 81% remained asymptomatic, and 89% showed either a reduction in lesion size or complete resolution.<sup>[19]</sup>

Early and accurate diagnosis is essential for the appropriate management of patients with CLMs. A standardised, multidisciplinary approach is required for diagnosis, follow-up, and treatment. Close collaboration among obstetricians, neonatologists, paediatric surgeons, and paediatric pulmonologists facilitates optimal management.

### Limitations

Detailed diagnostic test results for patients with prenatal diagnoses were unavailable. As a tertiary care centre, we frequently manage patients who have already undergone multiple investigations at other institutions, which may contribute to delays in diagnosis.

### CONCLUSION

CLMs are most commonly diagnosed during the prenatal period, and the majority of affected infants remain asymptomatic at birth. In asymptomatic prenatal cases, the first postnatal investigation should be chest X-ray, and routine CT scanning immediately after birth is not necessary. The diagnosis and management of CLM are not yet clearly defined; therefore, further prospective studies and stronger scientific evidence are required in this field. Management decisions should be based on an individual benefit–risk assessment.

### Statement

**Ethics Committee Approval:** The Istanbul Medipol Clinical Research Ethics Committee granted approval for this study (date: 22.05.2025, number: 644).

**Informed Consent:** Not applicable. This is a retrospective study based on anonymized patient data, and informed consent was not obtained by the institutional ethics committee.

**Conflict of Interest:** The authors declare that there is no conflict of interest.

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**Author Contributions:** Concept – EG, SCO; Design – SCO, SG; Supervision – SG; Results – SG, EG; Materials – YÖ; Data Collection and/or Processing – EG, YÖ; Analysis and/or Interpretation – YÖ, EG; Literature Search – EG; Writing – EG, SG; Critical Reviews – SG.

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