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ORIGINAL RESEARCH

Prenatal diagnostic accuracy and epidemiology of congenital lung malformations: A retrospective review of cases in a tertiary referral center in northern Finland in 2010–2020

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Abstract

Introduction: Our objective was to investigate the accuracy of prenatal diagnoses of congenital lung malformations (CLM) compared to postnatal diagnoses in a population in northern Finland and to estimate the birth prevalence of CLMs in the same population.

Material and Methods: A retrospective review of all CLM cases in a tertiary referral center, Oulu University Hospital, Finland, in 2010–2020. Data were collected from medical records. The final postnatal diagnosis was recorded as the pathologic-anatomic diagnosis, if available, and otherwise as the postnatal radiologic diagnosis.

Results: Our case series comprises 37 CLM cases. The prenatal detection rate of CLMs was 100%. The prenatal diagnosis was congenital pulmonary airway malformation (CPAM) in 34/37 cases (92%). The accuracy of prenatal CLM diagnoses was 60% compared to postnatal radiologic diagnoses and 51% compared to final postnatal diagnoses. Relative frequencies of different diagnoses in postnatally confirmed CLM cases were CPAM 47% (16/34 cases), bronchopulmonary sequestration (BPS) 15% (5/34), hybrid CPAM/BPS 15% (5/34), congenital lobar overinflation (CLO) 15% (5/34), bronchial atresia 6% (2/34), and bronchogenic cyst 3% (1/34). Postnatally confirmed cases of CPAM were more likely to have a higher CPAM-volume ratio at diagnosis ($p = 0.002$), a higher maximum CPAM-volume ratio during pregnancy ($p < 0.001$), macrocystic appearance on ultrasonography ($p = 0.026$), and mediastinal shift ($p < 0.001$) compared with the rest of the cases in this study. The prevalence of all CLMs combined was 3.71 cases per 10 000 live births. The prevalences of CPAM, BPS, hybrid CPAM/BPS, and CLO were 1.69, 0.56, 0.56, and 0.56 cases per 10 000 live births, respectively.

Conclusions: We found that all CLMs were detected prenatally, but almost half of the prenatal diagnoses were inaccurate compared to postnatal diagnoses. Most lesions were diagnosed prenatally as CPAM, but postnatally many of them turned out to be BPS, hybrid CPAM/BPS, or CLO. Postnatally confirmed CPAM cases were more likely to have a high CPAM-volume ratio, mediastinal shift, and macrocystic appearance on

Abbreviations: BMI, body mass index; BPS, bronchopulmonary sequestration; CLM, congenital lung malformation; CLO, congenital lobar overinflation; CPAM, congenital pulmonary airway malformation; CVR, CPAM-volume ratio; IQR, interquartile range; MRI, magnetic resonance imaging; OUH, Oulu University Hospital; PAD, pathologic-anatomic diagnosis.

prenatal ultrasonography compared with other CLMs. The prevalence of CLMs is still relatively poorly documented, but we provide new estimates in Finland.

KEYWORDS
pregnancy, prenatal diagnosis, prenatal, prevalence, respiratory system abnormalities/diagnostic imaging, retrospective studies, tomography, ultrasonography, X-Ray computed

1 | INTRODUCTION

Congenital lung malformations (CLM) are a group of pulmonary malformations that arise during fetal development. The most common diagnoses of this group include congenital pulmonary airway malformation (CPAM), bronchopulmonary sequestration (BPS), and congenital lobar overinflation (CLO, also known as congenital lobar emphysema).[1,2] All CLMs are rare, and estimates of their birth prevalence are relatively scarce. In recent years, the prevalence of CLMs has been estimated at 4–5 cases per 10 000 births, with CPAM considered to be the most prevalent CLM.[3,4]

CLMs are most commonly diagnosed prenatally using ultrasound and fetal magnetic resonance imaging (MRI). The prenatal differential diagnosis of a lung lesion is based on factors such as its location, echogenicity, and signal intensity, presence and size of cysts, and vascularization (i.e., whether the lesion has an abnormal feeding vessel).[5–7] CPAM may present as a homogeneous pulmonary lesion or with multiple cysts that may vary in size.[5,7] Bronchogenic cysts often present as a unilocular lesion in the mediastinum.[7] The presence of an abnormal arterial feeding vessel from systemic circulation into the lesion is a primary feature of BPS or a hybrid BPS lesion.[6,7] CLO is rarely diagnosed prenatally.[8,9]

The diagnostic accuracy of prenatal diagnoses of CLMs compared to postnatal diagnoses varies widely in previous studies.[2,5,10] Most often, CLMs are detected prenatally, but the specific pathology of the lesion and its vascularization can be misdiagnosed.[10] The presence of hybrid lesions of multiple pathologies further complicates prenatal diagnosis.[1,5] An accurate prenatal diagnosis may help improve prognostic evaluation and prenatal counseling.

The aim of this retrospective study is to evaluate the diagnostic accuracy of prenatal CLM diagnoses and estimate the prevalence of CLMs in a population in northern Finland. This information can be used to improve the diagnostic procedures for CLMs and to provide data on their prevalence in northern Europe. In addition, knowledge of the accuracy of prenatal diagnoses can be used in counseling parents regarding CLMs.

Key message

Congenital lung malformations are detected well prenatally, but the accuracy of prenatal diagnoses is only 51%–60% compared to postnatal diagnoses. The birth prevalence of all congenital lung malformations is estimated at 3.71 cases per 10 000 live births in northern Finland.

2 | MATERIAL AND METHODS

2.1 | Background

This study is a retrospective review of CLM cases treated at Oulu University Hospital (OUH) in Oulu, Finland. OUH functions as a regional tertiary referral center for complicated pregnancies and congenital anomalies in northern Finland, covering an area with a population of 737 000.[11] Almost all (99.7%–99.8%) pregnancies in Finland are monitored with regular visits to a maternal health clinic in primary care.[12] As part of the national maternal health program, a 1st trimester screening of the fetus is performed between 10+0 and 13+6 weeks of gestation, and an anomaly screening ultrasonography is offered to all mothers between 18+0 and 21+6 weeks of gestation. Primary and secondary care units in northern Finland refer pregnancies with suspicion of a congenital malformation to the fetal medicine unit at OUH.

2.2 | Data collection

Patients eligible for this study were those born between 1 January 2010 and 31 December 2020 who were diagnosed pre- and/or postnatally with a CLM and were monitored or treated at OUH before 1 January 2023, in addition to potential pregnancies that were terminated during the same period. Diagnoses included as CLMs in this study are CPAM, BPS, hybrid lesion of CPAM and BPS, CLO, bronchogenic cyst, and bronchial atresia. Cases were searched by ICD-10 codes (Q32.4 & Q33.0–Q33.9) from the hospital registries of OUH and by internal diagnosis codes from a database of the fetal medicine unit at OUH.

Cases were initially eligible if the medical records indicated that a CLM was diagnosed prenatally, postnatally, or both pre- and postnatally. However, cases were later excluded if the recorded CLM diagnosis was only prenatal and it had been discarded by a perinatologist before the gestational age of 30 weeks, or if the recorded CLM diagnosis was only postnatal (without prenatal indication) but the final postnatal diagnosis of the lesion was not a CLM.

Data were collected from both prenatal (maternal) and postnatal (pediatric) medical records. Data collected included maternal factors, prenatal ultrasonography findings, potential prenatal treatment,

neonatal health, neonatal treatment modalities, postnatal operative treatment, and postnatal diagnoses, including radiologic diagnoses and pathologic-anatomic diagnoses (PAD). Routine karyotyping was not offered to the mothers, and karyotype data were not collected in this study. Data were mainly collected from the medical records of OUH, but medical records of other hospitals were also available for patients who were prenatally diagnosed at OUH and later born and/or treated at another hospital, or vice versa.

CPAM-volume ratio (CVR) values were collected for prenatally suspected cases of CPAM in a standardized manner.[13] Values were obtained from medical records if they were described in the patient record. If CVR values were not available in the patient record, archived ultrasound images of the patient were reviewed by a perinatologist (T.K.) and used to retrospectively measure and calculate the CVR as part of the data collection. The equation used to calculate the CVR was: CVR = (lesion length × height × width × 0.52)/head circumference.[13]

The accuracy of prenatal diagnosis was evaluated by comparing the pre- and postnatal diagnoses in each case. Diagnoses were evaluated in order of reliability[10] from highest to lowest: postnatal PAD, postnatal radiologic diagnosis, and prenatal diagnosis (based on prenatal ultrasonography and fetal MRI). The final postnatal diagnosis was considered to be the postnatal PAD, if available, and otherwise the postnatal radiologic diagnosis.

2.3 | Statistical analysis

Symmetrically distributed numeric variables were described with mean and standard deviation, skewed numeric variables with median and interquartile range (IQR) (Q_1–Q_3), and categorical variables with frequencies and proportions (%). For numeric variables, the Mann–Whitney test was used to test for differences between two groups. For categorical variables, the N-1 chi-squared test and Fisher's exact test were used to test for differences between two groups.[14] Statistical significance was considered as a *p*-value below 0.05. Statistical analysis was performed using IBM SPSS Statistics for Windows 28.0.0.0 (IBM Corp., Armonk, NY, USA).

3 | RESULTS

3.1 | Case series

Our study describes a case series of 37 cases of CLM monitored or treated at OUH in 2010–2020. Four of these 37 cases (11%) were also prenatally monitored or postnatally treated at another hospital. During data collection, we initially identified 40 cases where a suspected CLM was pre- or postnatally monitored or treated at OUH in 2010–2020, but three of those cases were excluded from the final case series ($N = 37$) and are not included in any data further in the results. One of the excluded cases was a case of prenatally diagnosed CPAM that was deemed to be a diaphragmatic hernia after a fetal MRI study at the gestational age of 29 weeks. The second excluded case was diagnosed postnatally as CLO based on computed tomography, without any prenatal indication of a lung lesion. The patient underwent surgery, and the lesion was found to be congenital pulmonary lymphangiectasia, not a CLM, based on the PAD. The third excluded case was suspected prenatally to have a CPAM, but after birth, it became apparent that the patient also had a chromosome abnormality (Cat-eye syndrome) and several other congenital malformations, such as total anomalous pulmonary venous drainage of the heart and esophageal atresia. The patient died at the age of 5 weeks without postnatal imaging confirming the CPAM diagnosis, and as we did not have access to the obduction report, the case was excluded from the series.

3.2 | Prenatal data

All included cases ($N = 37$) progressed to labor, and there were no spontaneous or induced abortions. All postnatally confirmed CLM cases also had a prenatal CLM diagnosis, corresponding with a prenatal detection rate of 100% for all CLMs. Diagnosis of a CLM was given at a median gestational age of 22+0 weeks (IQR 21+3–22+5). This was recorded as the gestational age at which a CLM diagnosis was made by a perinatologist at a tertiary referral center. A primary care suspicion and referral of a CLM in our area lead to an ultrasonography exam by a perinatologist within three working days, with subsequent exams typically every 4 weeks in uncomplicated CLM cases. Prenatal ultrasonography was used in all cases and fetal MRI in 22 cases (59%). Maternal body mass index (BMI) data were available for 25/37 cases (68%), and the median BMI reported in the medical records at the time of diagnosis was 23.0 (IQR 20.5–27.0).

The use of fetal MRI in our center increased substantially over the studied time period as the availability of fetal MRI improved beginning in 2016. In 2010–2015, 3/16 cases (19%) underwent fetal MRI compared to 19/21 (90%) in 2016–2020. The diagnosis indicated by fetal MRI concurred with the primary ultrasonography diagnosis in 18/22 cases (82% of those that underwent fetal MRI), resolved a question of differential diagnosis that emerged in ultrasonography in 2/22 cases (9%), and provided a diagnosis that was not considered in ultrasonography in 2/22 cases (9%). The two resolved questions of differential diagnosis were whether a suprarenal lesion in ultrasonography was an extralobar BPS or splenomegaly, and whether an intrathoracic lesion found in ultrasonography was a CPAM or a diaphragmatic hernia. These cases turned out to be an extralobar BPS and a CPAM, respectively, in both fetal MRI and postnatally. Of the two cases where fetal MRI indicated a pathology that was not considered in ultrasonography, one was suspected to be a diaphragmatic hernia after the first ultrasonography study at OUH but was found to be a CPAM on fetal MRI, and CPAM was also the postnatal diagnosis. The other was suspected to be a left-sided CPAM based on ultrasonography but was diagnosed as a right lung hypoplasia on fetal MRI, with no lesion found on the left side.

Postnatally, this patient was diagnosed with right lung hypoplasia and esophageal atresia.

The most common prenatal diagnosis was CPAM in 34/37 cases (92%), followed by BPS in 2/37 cases (5%) and a bronchogenic cyst in 1/38 cases (3%). Cases of CPAM were almost equally divided between the left and right sides, with one bilateral case. The two prenatally diagnosed BPSs were intralobar in one case (right lower lobe) and extralobar in the other (attached to the left suprarenal gland).

The ultrasonographic appearance of the monitored CLMs regressed during pregnancy in 13/37 cases (35%), no significant change was observed in 11/37 (30%), a complete resolution was observed in 10/37 (27%), and the CLM increased in size in 3/37 cases (8%). Mediastinal shift was present in 21/37 cases (57%), and it resolved in 14/21 (67%) of those at a mean gestational age of 32 weeks (standard deviation 4.5 weeks). Polyhydramnios was observed in 4/37 cases (11%), and hydrops developed in 1/37 case (3%). Invasive intrauterine treatment was not needed in any case. Prenatal corticosteroid treatment was given in 5/37 (14%) cases.

Patient characteristics for different postnatally confirmed pathologies are described in Table 1. CVR values were acquired for all prenatally suspected cases of CPAM except for the one bilateral case ($N=33$). CVR was described in the patient record in 12/33 cases (36%) and measured retrospectively from archived ultrasound images during data collection in the remaining 21/33 (64%). The CVR at diagnosis was found to be the maximum CVR during pregnancy in 19/33 cases (58%). CVR at diagnosis had a median of 0.84 (IQR 0.46–1.50) and maximum CVR had a median of 1.15 (IQR 0.70–1.67).

Postnatally confirmed CPAM cases were more likely to have higher CVR at diagnosis ($p=0.002$) and higher maximum CVR during pregnancy ($p<0.001$), macrocystic appearance on ultrasonography ($p=0.026$), and mediastinal shift ($p<0.001$) compared with the rest of the cases in this study. No other pathology, besides CPAM, showed a statistically significant difference in any prenatal variable compared with the rest of the case series (Table 1).

3.3 | Postnatal data

All included cases resulted in live birth with a median gestational age at birth of 39+1 weeks (IQR 38+3–39+6). All 37 children (100%) were alive at the time of data collection (median age 6.6 years, IQR 4.6–10.2). The most common postnatal radiologic CLM diagnosis was CPAM in 19/37 cases (51%) (Table 2). The imaging modality used to assign the postnatal radiologic diagnosis was computed tomography in 35/37 cases (95%), native radiography in 1/37 cases (3%), and ultrasonography in 1/37 cases (3%).

PAD was obtained for all patients who had a surgery for their CLM before the time of data collection in this study (30/37, 81%). Median age at resection was 12 months (IQR 7–18). The most common PAD was CPAM in 15/30 cases (50% of all PADs), followed by BPS in 5/30 cases (17%), and hybrid CPAM/BPS in 4/30 cases (13%). The cases with only native radiography and ultrasonography as postnatal imaging modalities were among those confirmed with PADs.

3.4 | Diagnostic accuracy

The overall diagnostic accuracy of the prenatal CLM diagnoses was 60% (22/37 cases) compared to postnatal radiologic diagnoses and 51% (19/37) compared to final postnatal diagnoses. The final postnatal diagnosis was considered to be the postnatal PAD, if available, and otherwise the postnatal radiologic diagnosis. The postnatal radiologic diagnoses agreed with the available PADs in 87% of the cases (26/30). The radiology report of fetal MRI studies included the final

TABLE 1 Patient characteristics in different congenital lung malformations based on the final postnatal diagnosis.

Patient characteristic	All included cases ($N=37$)	Postnatal CPAM ($N=16$)	Postnatal BPS or hybrid CPAM/BPS ($N=10$)	Postnatal CLO ($N=5$)
Lesion resolution during pregnancy, *n* (%)	10 (27)	3 (19) $p=0.322$	2 (20) $p=0.558$	3 (60) $p=0.074$
Microcystic appearance on ultrasonography, *n* (%)	17 (46)	**4 (25)** $p=0.026$	5 (50) $p=0.763$	4 (80) $p=0.100$
Mediastinal shift present during pregnancy, *n* (%)	21 (57)	**14 (88)** $p<0.001$	5 (50) $p=0.614$	1 (20) $p=0.074$
CVR at diagnosis, median (IQR)[a]	0.53 (0.27–0.84)	**0.84 (0.46–1.50)** $p=0.002$	0.53 (0.19–0.62) $p=0.467$	0.38 (0.27–0.54) $p=0.238$
Maximum CVR during pregnancy, median (IQR)[a]	0.68 (0.42–1.15)	**1.15 (0.70–1.67)** $p<0.001$	0.64 (0.22–0.79) $p=0.415$	0.56 (0.42–0.69) $p=0.292$
Gestational age in weeks at maximum CVR, median (IQR)[a]	22.0 (21–25)	23 (21–25) $p=0.700$	22 (22–23) $p=0.653$	24 (21–25) $p=0.939$

Note: Bolded values indicate a statistically significant ($p<0.05$) difference between a group and the rest of the cases in this study in statistical comparisons performed using the $N-1$ chi-squared test and Fisher's exact test for categorical variables, and the Mann–Whitney test for numeric variables.

Abbreviations: BPS, bronchopulmonary sequestration; CLO, congenital lobar overinflation; CPAM, congenital pulmonary airway malformation; CVR, CPAM-volume ratio; IQR, interquartile range.

[a]CVR data were available for prenatally suspected unilateral CPAM cases ($N=33$).

TABLE 2 Prenatal congenital lung malformation diagnoses and their corresponding postnatal diagnoses based on different diagnostic modalities.

Prenatal	Postnatal		
USG diagnosis (*N*)	Radiologic diagnosis (*N*)	PAD (*N*)	Final diagnosis (*N*)[a]
CPAM (34)	CPAM (19)	CPAM (15)	CPAM (16)
	BPS (2)	BPS (3)	BPS (3)
	Hybrid CPAM/BPS (6)	Hybrid CPAM/BPS (4)	Hybrid CPAM/BPS (5)
	CLO (2)	CLO (3)	CLO (5)
	Bronchial atresia (2)	Bronchial atresia (1)	Bronchial atresia (2)
	Lung hypoplasia (1)	Lung hypoplasia (1)	Lung hypoplasia (1)
	No lung defect (2)	*Not available* (7)	No lung defect (2)
BPS (2)	BPS (2)	BPS (2)	BPS (2)
BC (1)	BC (1)	BC (1)	BC (1)

Abbreviations: BC, bronchogenic cyst; BPS, bronchopulmonary sequestration; CLO, congenital lobar overinflation; CPAM, congenital pulmonary airway malformation; PAD, pathologic-anatomic diagnosis; USG, ultrasonography.

[a]Final diagnosis listed as PAD if available and otherwise as the postnatal radiologic diagnosis.

postnatal diagnosis as one of the possible differential diagnoses in 15/22 cases (68% of all fetal MRI cases).

The accuracy of diagnoses did not change significantly during the studied time period 2010–2020 ($p=0.902$), cases with fetal MRI were not more likely to have an accurate diagnosis compared to cases without MRI (50% vs. 60%, $p=0.549$), and maternal BMI was not associated with the accuracy of prenatal diagnosis ($p=0.978$). A feeding vessel related to BPS was present in 10 cases according to the final postnatal diagnosis, but the feeding vessel was detected prenatally in only two of these cases (20%). No feeding vessels were missed by postnatal imaging compared to PAD (9/9 detected).

Considering the different prenatally suspected CLMs, a prenatally suspected CPAM ($N=34$) was confirmed as a CPAM on the postnatal radiologic diagnosis in 19 (56%) of cases and confirmed as a CPAM in the final postnatal diagnosis in 16 (47%) of cases. For prenatally diagnosed BPS ($N=2$) and bronchogenic cyst ($N=1$), the accuracy of prenatal ultrasonography was 100% (2/2 and 1/1 cases, respectively) compared to both the postnatal radiologic diagnosis and the final postnatal diagnosis. There were no other prenatally suspected diagnoses in the included cases.

3.5 | Epidemiology

Birth prevalences for different postnatally confirmed CLM diagnoses are described in Table 3. Prevalences were not provided separately for diagnoses with three or fewer cases identified in this study. One case was excluded from the prevalence calculations because it belonged to the referral area (population) of another tertiary center at the time of birth ($N=36$). During the time period 2010–2020, a total of 88 839 children were born alive in the area that is covered by OUH as a tertiary referral center.[11] This was the number of live births used to calculate the birth prevalences. The relative frequencies of final diagnoses among all postnatally confirmed CLM cases in our case series were CPAM 47% (16/34 cases), BPS 15% (5/34), hybrid CPAM/BPS 15% (5/34), CLO 15% (5/34), bronchial atresia 6% (2/34), and bronchogenic cyst 3% (1/34).

TABLE 3 Birth prevalences of different congenital lung malformation diagnoses in the tertiary referral area of Oulu University Hospital in 2010–2020 based on postnatally confirmed diagnoses, except for the prevalence of prenatally suspected CLMs.

Diagnosis	Cases (*N*)	Prevalence per 10 000 live births
Prenatally suspected CLMs	36	4.05
All postnatally confirmed CLMs combined	33	3.71
CPAM	15	1.69
BPS	5	0.56
Hybrid CPAM/BPS	5	0.56
CLO	5	0.56

Note: One CPAM case, which was registered within the referral area of another tertiary center at birth, was excluded from the calculations ($N=36$). Prevalences were calculated using the number of live births (88 839) in the referral area of Oulu University Hospital in 2010–2020, as reported by Official Statistics of Finland.

Abbreviations: BPS, bronchopulmonary sequestration; CLM, congenital lung malformation; CLO, congenital lobar overinflation; CPAM, congenital pulmonary airway malformation.

4 | DISCUSSION

We report a retrospective study of all CLM cases in our tertiary referral center over a period of 11 years. We investigated the

diagnostic accuracy of prenatal CLM diagnoses and found that CLMs were detected prenatally in all cases, but almost half of the prenatal diagnoses were inaccurate compared to the postnatally confirmed diagnoses. The prenatally suspected pathology was CPAM in all but three cases, but these lesions were postnatally confirmed as CPAM in only approximately half of the cases. The prenatally misdiagnosed cases most commonly included a feeding vessel that was not detected, or they were postnatally found to be CLO instead of CPAM. We found that postnatally confirmed CPAM cases were more likely to have high CVR, mediastinal shift, and macrocystic appearance on ultrasonography than the rest of the case series. Using nationally collected birth statistics, we also calculated birth prevalences for all CLMs combined as well as for the most common CLM diagnoses in a population in northern Finland.

The overall diagnostic accuracy of prenatal CLM diagnoses in our study was 51% when compared to the final postnatal diagnoses. This corresponds well with previous studies where similarly measured accuracies have ranged from 35% to 74%.[5,15,16] The wide variation in the reported diagnostic accuracies raises the question of whether some factors that are associated with the accuracy could be identified. We investigated whether the year of birth of the child (i.e. improvement of ultrasound technology over time), maternal BMI (i.e., more abdominal adipose tissue that could hinder the ultrasonography study), or the use of fetal MRI were associated with the accuracy of prenatal diagnosis, but no such associations were found. Considering the impact of fetal MRI, patient selection should not be a significant confounding factor in our study, since the availability and use of fetal MRI (in CLM cases) in our center increased from very low to an almost universal level beginning in 2016, but neither the year of birth nor use of fetal MRI were still associated with improved diagnostic accuracy in our results. Considering the effect of BMI, it should be noted that our BMI data were quite limited. The prenatal detection rate of postnatally confirmed CLMs in our study was 100%, which is similar to detection rates of 83%–94% reported in previous studies.[17,18]

Prenatally suspected CPAMs comprised most cases in our study, and all prenatal misdiagnoses in our data were in this group. The prenatal suspicion of CPAM was confirmed by the final postnatal diagnosis of CPAM in 47% of cases, which is lower than in previous studies, where diagnostic accuracies of 62%–89% have been reported for prenatal diagnosis of CPAM.[2,10,15,18] The prenatal misdiagnoses of CPAM in our study were most commonly found to have a (missed) feeding vessel (i.e. final diagnosis of BPS or hybrid CPAM/BPS) or to be CLO. Our prenatal detection rate of abnormal feeding vessels (20%) was considerably lower compared with the detection rates in other studies within the last 10 years (49%–88%).[10,16,18,19] We did not recognize any factors explaining this large difference. We did note that the ultrasonography reports that were examined during data collection in this study often mentioned the absence of abnormal vascularization, indicating that feeding vessels were actively searched for by perinatologists in our center. These findings suggest that BPS should not be ruled out in ultrasonography even if no feeding vessel is observed in ultrasonography. Other ultrasonographic features may aid in differential diagnosis.

We found that high CVR, mediastinal shift, and macrocystic appearance on ultrasonography were associated with postnatally confirmed CPAM cases. This would suggest that small lung lesions with microcystic features on prenatal ultrasonography are especially important to consider for differential diagnoses other than CPAM. All five CLO cases in our study were detected prenatally, but all of them were prenatally misdiagnosed as CPAM. A large register study previously found somewhat similarly that CLO is often misdiagnosed, most commonly as CPAM, or not detected prenatally.[8] Our five CLO cases were described as microcystic and without mediastinal shift on prenatal ultrasonography more often than the rest of the case series, but these differences were not statistically significant. However, the homogeneous and echogenic (i.e., microcystic) appearance of CLO on ultrasonography has also been reported in previous literature.[9,20] It seems that although CLO is difficult to diagnose correctly prenatally, it may present more often as microcystic (homogeneous) and without mediastinal shift on ultrasonography compared with other CLMs.

Prevalence estimates for CLMs are historically relatively scarce in the literature, but some recent estimates are available for the most common diagnosis of CPAM and for CLMs in general. Older estimates are less relevant because the prevalence of CLMs has been increasing considerably with time.[17,21] A European registry estimates the prevalence of CPAM at 1.26 per 10000 live births in 2020, and the data show an increasing trend over time.[22] A universal ultrasonography program study in Hong Kong estimated the prevalence of CPAM at 1.39 per 10000 live births in 2009–2014,[17] a large register study in China estimated the prevalence at 1.21 per 10000 live and still births in 2010–2019,[21] and a study in Brazil estimated the prevalence substantially higher at 5.05 per 10000 births in 1990–2010.[4] For CLMs in general, one study in the United Kingdom reports a prevalence of 4.15 per 10000 births in 2008–2012.[3] The prevalence of CPAM and CLM in our data—1.69 and 3.71 cases per 10000 births, respectively—corresponds relatively well with the values reported in previous studies. We believe a factor strengthening the reliability of our estimates, despite the limited size of our case series, is the almost universal use of primary care maternal health clinics, which refer suspected congenital malformations to our tertiary center in our population. The number of live births used in our calculations can also be considered reliable as all births in Finland are recorded for national statistics based on the referral areas of tertiary centers. Overall, our findings suggest that the prevalence of CPAM and CLM in northern Finland during the last decade is similar to previously reported estimates in other populations. The prevalence of all CLMs seems to be around 4 per 10000 births according to our findings and previous literature.

Considering the relative frequencies of different CLM diagnoses, our data are in relatively good agreement with previous studies. In two large studies from the United States, the frequencies of the most common pathologically confirmed diagnoses of patients that underwent resection for a CLM were CPAM 44%–47%, BPS 24%–35%,

hybrid CPAM/BPS 9%–21%, CLO 8%–11%, and bronchogenic cysts 1%–7%.[1,18] Our data and the other two studies agree that CPAM accounts for slightly less than half of all CLM cases and CLO for around 10%, but BPS and hybrid CPAM/BPS were more common in the previous studies compared with our data. However, the inclusion criterion of having undergone resection in the other studies represents a screening factor that could significantly influence the frequencies. We believe that more data are needed on the epidemiology and relative frequencies of CLMs in unscreened study populations.

Strengths of our study include comprehensive diagnostic data for all included cases and reliable inclusion of all CLM cases in our tertiary referral area due to an almost universally used system of primary care maternal health clinics. It is unlikely that cases of suspected CLM in the tertiary referral area of our center were not referred to the fetal medicine unit at OUH. The Finnish national maternal health program also resulted in good comparability between our cases, as gestational age at diagnosis was similar for all cases.

Limitations of our study include the relatively small number of cases and limited data on some prenatal factors. Even though our study represents an 11-year record of all CLM cases in our center, the resulting case series is limited to 37 patients. This may affect both the detection of potential differences in prenatal factors between pathologies and our prevalence estimates. Data on some prenatal factors, such as maternal BMI, were not available for all cases in our study. A better coverage of all prenatal factors might have revealed the diagnostic significance of some variables that showed no prominence in this study.

5 | CONCLUSION

Our study reports a retrospective review of all CLM cases in a tertiary referral center in northern Finland in 2010–2020. We found that all postnatally confirmed CLM lesions were detected by prenatal ultrasonography, but almost half of all prenatal CLM diagnoses in our case series were inaccurate compared to the final postnatal diagnoses. Most cases were prenatally suspected to be CPAM, but many of these were postnatally confirmed to be BPS, hybrid CPAM/BPS, or CLO. Postnatally confirmed CPAM cases were more likely to have high CVR, mediastinal shift, and macrocystic appearance on prenatal ultrasonography compared with other CLMs. Based on these findings, we recommend considering a differential diagnosis other than CPAM, especially in smaller lesions with microcystic appearance. Further research is needed to identify possible factors associated with certain CLMs and their diagnostic accuracy.

AUTHOR CONTRIBUTIONS

Topias Puumalainen was involved in conceptualization, methodology, data collection, analysis, writing, and reviewing. Tuomas Kauppinen was involved in conceptualization, methodology, data collection, and reviewing. Hilkka Nikkinen was involved in conceptualization, methodology, and reviewing. All authors read and approved the final manuscript.

CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

ETHICS STATEMENT

This study was approved by the Northern Ostrobothnia Hospital District (142/2022) on August 12, 2022. Separate ethical approval was not required, as only patient records were used in data collection.

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