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Original Article

Spectrum of Congenital Lung Malformations in Children: Experience from A Tertiary Care Center

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ABSTRACT

Aim: A variety of developmental abnormalities of the tracheobronchial tree and pulmonary parenchyma are found in the newborn. There is limited data available on their presentation and clinical course from third world countries.

Materials and Methods: A retrospective review of the medical records of children with congenital lung malformations was conducted at our tertiary care hospital from June, 2014 to June, 2018.

Results: This study included 30 patients with 12 males and 18 females. The mean age at diagnosis was 13 months (range 1 month to 4 years). Congenital pulmonary airway malformation was the most common malformation present in 18 (60%) patients, followed by congenital lobar emphysema in 4 (13.3%), bronchogenic cyst in 3 (10%), bronchopulmonary sequestration in 2 (6.7%), bronchial atresia in 2 (6.7%) and pleuropulmonary blastoma in 1 (3.3%) patient. The most common presentation was recurrent upper respiratory tract infection (53.3%). Lobectomy was the most common surgical procedure performed in 86.6% of the patients, followed by excision of cyst in 10% of the patients. The post-operative complication rate was 30% with an overall survival rate of 93.3%. The average follow-up duration was 9 months.

Conclusion: The predominant lung malformation in this study was congenital pulmonary airway malformation. Recurrent respiratory tract infection with or without respiratory distress was the most common mode of presentation. Health education to raise awareness and emphasis on antenatal ultrasonography can avoid inordinate delays in diagnosis and treatment. Surgery is curative with good long-term outcomes.

Keywords: Congenital pulmonary airway malformation, bronchogenic cyst, pulmonary sequestration, congenital lobar emphysema

Introduction

Congenital lung malformations (CLMs) are a group of rare lung abnormalities affecting the airways, parenchyma, and vasculature. CLMs include congenital pulmonary airway malformations (CPAM), pulmonary sequestrations (PS), bronchogenic cysts, congenital lobar emphysema (CLE) and bronchial atresia (1). Many cases have overlapping features with CPAM like changes in an area of sequestered lung referred to as hybrid lesions (2,3). They are rare congenital malformations with a cumulative incidence of 30-42 cases per 100,000 individuals (4).

Due to the rarity of these malformations and the paucity of literature from the developing world, this study was conducted to share our experiences from a tertiary care center, which is the only center in the region catering to the treatment of such anomalies.

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Materials and Methods

This was a retrospective data review, conducted at a tertiary care hospital in northern India. The medical records of those patients diagnosed with CLM from June, 2014 to June, 2018 were evaluated. This study was undertaken after due approval by the Institutional Review Board of Sher-I-Kashmir Institute of Medical Sciences (approval no: 80/2018, date: 18.06.2018). The study population included children diagnosed as having CLM based on their clinical, radiological, and pathological features. We excluded syndromic children and those with major cardiac malformations. Data on their demographic profiles such as their age and gender, clinical presentation, baseline investigations, chest imaging studies (X-ray and computed tomography), hospital admissions, surgical interventions, surgical outcomes and follow-ups were retrieved. Any other relevant investigations such as bronchoscopy or dye study findings were also noted. Surgical details which included the timings of their surgical intervention, the type of surgery, the duration of the operation, intraoperative findings, and their intraoperative and postoperative complications were recorded. The CLMs studied included CPAM, CLE, bronchopulmonary sequestration, bronchial atresia, bronchogenic cysts and pleuropulmonary blastoma.

Statistical Analysis

The final data was entered into Microsoft Excel. Data analysis was performed using SPSS software (SPSS Inc., Chicago, IL, USA). The values of various parameters are presented as mean ± standard deviation, in absolute numbers or as percentages.

Results

Between June, 2014 and June, 2018, we studied 30 patients with CLMs in our hospital. The malformations were CPAM 18 (60%), CLE 4 (13.3%), bronchogenic cyst 3 (10%), sequestrations 2 (6.7%), bronchial atresia 2 (6.7%) and pleuropulmonary blastoma 1 (3.3%). The baseline characteristics of the patients are listed in Table I.

Most of the patients presented with respiratory tract infection/pneumonias (53.3%), followed by fever (26.6%), difficulty in breathing (23.3%) and pneumothorax (13%). Antenatal diagnosis was made on ultrasonography, which showed a cystic lesion in the chest in 3 (10%) patients. X-ray and computed tomography (CT) chest was performed in all patients (Figure 1). The most common finding on chest radiograph was cystic lucency in 83% of the patients, 50% of the patients had the presence of infiltrates, 33.3% patients had mediastinal shift to the opposite side, pneumothorax was identified in 13.3% of the patients and 6.6% of the patients had atelectasis. Chest ultrasound indicated a significant finding in 73.3% of the patients. Cystic lesion was the most common finding present in 36.6% of the patients, followed by consolidation in 33.3% of the patients, pleural effusion in 33.3%, pneumothorax in 13.3% and atelectasis was noted in 10% of the patients. CT of the thorax indicated abnormalities in lung parenchyma which included cystic lesions (73.3%), consolidation (33.3%), pleural effusion

Table I. Base line characteristics of the study population (n=30)

Characteristic	n (%) or mean
Age at presentation (months)*	13 (1.48)
Antenatal diagnosis	3 (10)
Male gender	12 (40)
Presenting symptom	
Asymptomatic	3 (10)
Symptomatic	27 (90)
Type of malformation	
Congenital pulmonary airway malformation	18 (60)
Congenital lobar emphysema	4 (13.3)
Pulmonary sequestration	2 (6.7)
Bronchogenic cyst	3 (10)
Bronchial atresia	2 (6.7)
Pleuropulmonary blastoma	1 (3.3)
Operation/Surgical resection done	30 (100)
Duration of follow-up (months)*	9 (3.36)
Associated malformations	8 (26.6)

*Mean and range

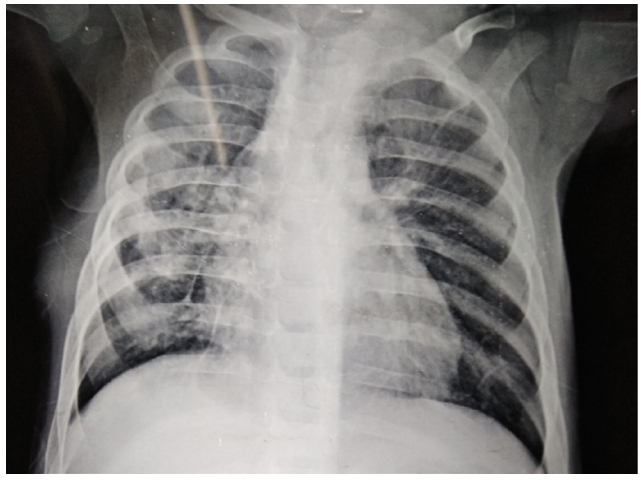

Figure 1. X-ray chest showing cystic lucency right upper lobe in a patient with right upper lobe congenital pulmonary airway malformation

(26.7%), pneumothorax (13.3%), bronchiectasis (10%) and atelectasis (10%) (Figure 2). The diagnoses of all of the patients was preoperatively established for CPAM, CLE, and bronchial atresia using different radiological investigations, particularly computed tomography, whereas the diagnoses of 50% of the patients with sequestration and a patient with pleuropulmonary blastoma were confirmed after histopathological examination.

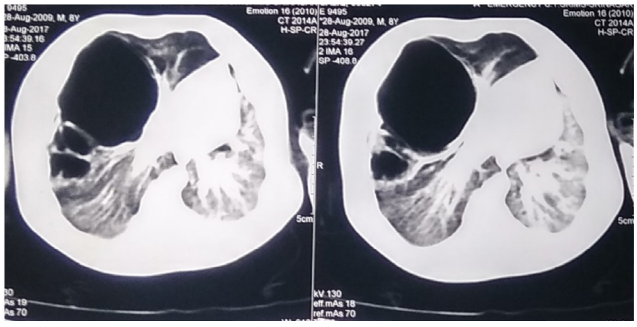

Figure 2. CT chest showing large cyst in right upper lobe congenital pulmonary airway malformation

Surgical procedures consisted of lobectomy (86.6%), cystectomy (10%), and others (3.3%) (Figure 3). Postoperative complications were pneumothorax (13.3%), sepsis (10%) and wound infection (6.6%). The clinical profiles, demography, and the outcomes of various CLMs are shown in Table II.

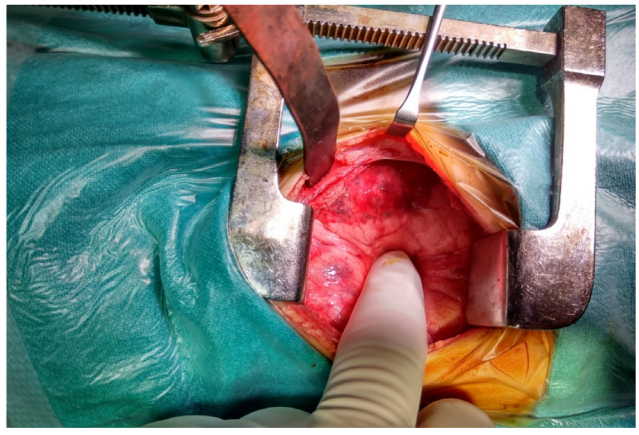

Figure 3. Intraoperative picture in a patient with right upper lobe congenital pulmonary airway malformation

Table II. Clinical profile, demographic characteristics, and outcome of various congenital lung malformations

Characteristic	CPAM (n=18)	CLE (n=4)	Sequestration (n=2)	Bronchogenic cyst (n=3)	Bronchial atresia (n=2)
Location, n (%)					
Right upper lobe	6 (33.3)	3 (75)	0	0	0
Right middle lobe	4 (22.2)	0	0	0	1 (50)
Right lower lobe	5 (27.7)	0	0	0	0
Left upper lobe	2 (11.1)	1 (25)	0	2 (66.6)	0
Left lower lobe	1 (5.5)	0	2 (100)	1 (33.3)	1 (50)
Age at diagnosis (mo)*	12.4 (2.48)	2.3 (2.3)	24	4.6 (1.9)	19 (14.24)
Age at surgery (mo)*	12.4 (2.48)	2.3 (2.3)	24	4.67 (1.9)	19 (14.24)
Male gender, n (%)	8 (44.4)	2 (50)	1 (50)	0	1 (50)
Presenting symptom, n (%)					
Fever	4 (22.2)	1 (25)	2 (100)	1 (33.3)	0
Recurrent chest infections	11 (61.1)	1 (25)	1 (50)	1 (33.3)	1 (50)
Fast breathing	4 (22.1)	2 (50)	0	0	0
Pneumothorax	4 (22.1)	0	0	0	0
Chest X-ray findings, n (%)					
Cystic lucencies	18 (100)	2 (50)	1 (50)	3 (100)	1 (50)
Mediastinal shift	7 (38.8)	2 (50)	0	1 (33.3)	0
Presence of infiltrates	10 (55.5)	1 (25)	1 (50)	2 (66.6)	1 (50)
Pneumothorax	4 (22.2)	0	0	0	0
Atelectasis	4 (22.2)	1 (25)	0	2 (66.6)	1 (50)
Postoperative complications, n (%)	5 (27.7)	1 (25)	1 (50)	0	1 (50)
Mortality, n (%)	0	1 (25)	0	0	0

*Mean and range, CPAM: Congenital pulmonary malformation, CLE: Congenital lobar emphysema

One operated case of pleuropulmonary blastoma died one year after surgery during follow-up by the medical oncology department. One case of Bronchogenic Cyst died 10 days after surgery (5 days after discharge from the hospital).

Discussion

Congenital pulmonary malformations are rare lesions which form during the embryological development phase of the lungs. The rates of diagnosis for these rare malformations in both the prenatal period and adulthood have increased because of advances in imaging methods (5).

In our study, CPAM was the most common type of CLM which is similar to previous reports (6-8). Bronchogenic cyst and CLE, however, have also been reported as major CPMs in some studies (9,10). We observed that the age distribution in our patients was 1 month to 4 years with a mean age of 13 months. The mean age at the time of diagnosis has been reported as being 13.1 months (11). CPAM and CLE in our study presented at an earlier age than PS. Patients with CPAM and CLE may present with symptoms earlier than patients with PS as the lesions of CPAM and CLE may compress the tracheobronchial tree, which can cause earlier symptom presentation (12).

On reviewing the clinical presentations of these lesions, respiratory tract infection/pneumonia (53.3%), fever (26.6%), difficulty in breathing (23.3%), and pneumothorax (13%) were the most commonly presenting symptoms. These findings are consistent with the reports of Jamero et al. (13). It has been observed that neonates and infants generally present with signs of respiratory distress (dyspnea, cyanosis) while older children mainly present with signs of pulmonary infection (fever, cough) (9,14).

Chest imaging is the cornerstone of diagnosis. Chest X-ray is usually the first line of investigation, and its findings vary with the type of malformation. CT is the gold-standard modality for the evaluation of anatomical structural defects and to define the anomalous blood supply (15). Large cystic lesions may present with changes affecting an entire hemi thorax, over inflation, mediastinal shift, and atelectasis (15). The location of the lung cyst in our study was in line with previously published reports (13). CPAM predominantly involved the right lung. The right upper lobe and the right lower lobe were more commonly involved. Bronchogenic cyst mostly involved the right lung. The right upper lobe was the most involved lobe. Congenital Lobar Emphysema involved the left lung in all cases. The left upper lobe was the most involved lobe, whereas PS involved the left lower lobe in all the cases.

All patients of CLM underwent some form of surgery in our study after preoperative optimization. While the decision making for children with symptomatic CLMs is reasonably straight forward, namely resection of the abnormality, there is ongoing debate regarding the need for and the timing of surgery in children with asymptomatic lesions. For those who support surgical intervention, the safety of pulmonary resection in infants and children is no longer a source of debate as its outcomes are generally good. Complication rates after surgery range between 6% and 9% and are mostly related to prolonged air leak (16). Mortality is a very rare occurrence in experienced hands and is thus not a significant outcome measure. The major surgical procedure in the present study was lobectomy and the main postoperative complications were pneumothorax, sepsis, and/or wound infection, similar to previous studies (8,17).

In the present study, there was an overall survival rate of 93.33%. There were 2 deaths. One operated case of pleuropulmonary blastoma died 1 year after surgery during follow-up by the medical oncology department. Pleuropulmonary blastoma can mimic CPAM. There should be a high index of suspicion in order to diagnose this rare and aggressive tumor (18). One case of bronchogenic cyst died 10 days after surgery (5 days after discharge from the hospital), probably due to aspiration.

Study Limitations

This study had several limitations. It was retrospective in design. The sample size was small due to the rarity of these malformations, which reduced the power of this study. Also, there was no data on lung function tests or long-term neurodevelopment outcomes. This study, nevertheless, gives an insight into these rare malformations.

Conclusion

In conclusion, CPAM was the most common CLM in our study. Recurrent respiratory tract infection with or without respiratory distress was the most common mode of presentation. Early detection of these relatively rare malformations will significantly improve the outcomes for such children. Surgery is the best modality of treatment with good outcome. Further studies from developing countries, with information regarding long-term follow-ups including lung function assessments, are needed.

Ethics

Ethics Committee Approval: The ethical approval was obtained from the Institutional Review Board of Sher-I-Kashmir Institute of Medical Sciences (approval no: 80/2018, date: 18.06.2018).

Informed Consent: Informed consent was obtained.

Peer-review: Internally and externally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: G.N.M., W.J.S., A.S., N.A.B., A.A.B., R.H., S.K.A., Concept: S.M.A., G.N.M., W.J.S., A.S., N.A.B., A.A.B., R.H., S.K.A., Design: G.N.M., W.J.S., A.S., N.A.B., A.A.B., R.H., S.K.A., Data Collection or Processing: S.M.A., W.J.S., A.S., Analysis or Interpretation: S.M.A., G.N.M., W.J.S., A.S., N.A.B., A.A.B., R.H., S.K.A., Literature Search: S.M.A., W.J.S., A.S., Writing: S.M.A., W.J.S., A.S., S.K.A.

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