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ORIGINAL ARTICLE

Clinical Presentation and Outcome of Congenital Thoracic Malformations in Children Beyond Neonatal Period

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ABSTRACT

Objective: To describe the clinical spectrum of presentation and outcome of children with congenital thoracic malformations beyond neonatal age.

Methodology: Cross sectional study conducted at inpatient department of National institute of child health from Jan-Dec 2021. All patients hospitalized in study duration with diagnosis of congenital thoracic formations from 1 month till 12 years of age were enrolled. History, examination, laboratory tests, treatment and outcome were recorded.

Results: Total 44 children were enrolled with mean age of 7.7⁺10.8 months. Common congenital thoracic malformations were congenital diaphragmatic hernia 24(54.5%), congenital cystic adenomatoid malformation 6(13.6%), congenital lobar emphysema5(11.4%) and eventration of diaphragm 4(9.1%). Three (6.9%) patients had associated cardiac anomalies. Surgical intervention was done in 33 (75%) patients with a postoperative survival rate of 95.4%.

Conclusion:
Congenital diaphragmatic hernia and cystic adenomatoid malformation are common congenital thoracic malformations that present beyond neonatal age and are associated with good postsurgical outcome.

Keywords: congenital diaphragmatic hernia, congenital cystic adenomatoid malformation, beyond neonate

INTRODUCTION

Congenital thoracic malformations (CTM) are very rare developmental anomalies and in international literature CTM is being used to describe congenital lung malformations and diaphragmatic abnormalities and cardiac malformations are studied sparately.[1,2] Estimated annual incidence of congenital lung malformations (CLM) is 30-42 per 100,000 live births and of congenital diaphragmatic abnormalities of which most common is congenital diaphragmatic hernia (CDH) is 1 in 3000 live births.[1,3]

CLM include variety of developmental problems of pulmonary system and include congenital pulmonary airway malformation (CPAM), bronchial cysts, congenital segmental or lobar emphysema, congenital cystic adenomatoid malformation (CCAM), bronchopulmonary sequestration (BPS) and vascular malformations.[4]

CDH is the birth defect in diaphragm with herniation of intra-abdominal viscera in chest cavity causing pulmonary hypoplasia and pulmonary hypertension which are main determines of morbidity and mortality and in some cases, it is also associated with other anomalies of genitourinary, cardiovascular and brain.[5,6] It has three types, posterolateral Boc dalek hernia, anterior Morgagni hernia and hiatal hernia. [3,4]

Course and outcome of CLM and CDH has variable natural history and depends upon size of lesion and underlying pathology.[7] In large lesion, it may put pressure on surrounding structures and may lead to severe respiratory distress, heart failure, non-immune fetal hydrops, pulmonary hypoplasia respiratory failure and pulmonary hypertension.[7] Approximately 1 in 5 cases of CLM are diagnosed after neonatal period incidentally due to respiratory distress.[8]

Optimal timing for surgery is controversial. Early surgery may have benefit of better compensatory growth of lung and prevention of pulmonary hypertension but few authors recommend late surgery with good prognosis.[7] Symptomatic infants in neonatal or antenatal period are benefitted by early surgery in neonatal period while in asymptomatic children, treatment varies from case to case according to the degree of severity.[8]

Despite of improvement in antenatal detection, only 60% CDH are diagnosed antenatally and it has poor outcome as compared to post-natal diagnosed CDH as smaller defects are more likely to be missed on antenatal imaging.[9] The overall survival in CDH is around 80% in western countries but long term functional disabilities may persist.[10] In associated anomalies, 15 % may have associated congenital heart defects and may be related to worsened outcome.[11] Common presentation is in newborn period with severe respiratory distress and mortality is around 30% in newborn period and morbidity is significantly high due to long term problems in lung functions and associated anomalies.[12] Around 10 % are associated with chromosomal abnormalities including defects in chromosomal number, rearrangements and deletions identified by karyotyping , microarrays and FISH probing. [13]

Congenital thoracic malformations are rare and little local data is available beyond neonatal age. We conducted this study with objective to describe the clinical spectrum of presentation and outcome of children with congenital thoracic malformations beyond neonatal age in order to provide local data in children presenting after neonatal age with these malformations. We expect our data will help in making future workup and management strategies for early diagnosis and improving the outcome.

METHODOLOGY

This prospective cross-sectional study was conducted during January to December 2021 in all three medical units of National Institute of Child Health (NICH) Karachi. NICH is a 500 bedded only public sector tertiary care referral center in Sindh province with facility of pediatric surgery. All patients admitted in medical wards of NICH with the diagnosis of congenital thoracic malformations from 1 month till 12 years of age during the study period were enrolled after taking informed consent from parents.

Detailed medical history and examination were performed and documented. Chest Xray and CT scan findings were recorded. All children received routine hospital care according to hospital protocol and details were recorded in the form of medical and surgical management. Outcome was recorded in form of discharge, left against medical advice or death. Data was analyzed using SPSS version 25.

RESULTS

Total 44 children were enrolled with mean age of 7.7⁺10.8 SD, range (1-48) months out of which 28(63.6%) were male and 16(36.4%) were female. Mean weight was 5.3⁺2.9 kg. Most common clinical presentation was difficulty in breathing present in all patients and others clinical features are explained in table 1.

Table 1: Clinical features in children with congenital thoracic malformations N=44

Clinical feature	Number (percentage)
Respiratory distress	44(100%)
Cough	35(79.5%)
Feeding difficulty	21(47.7%)
Failure to thrive	19(43.2%)
Chest deformity	10(32.7%)
Wheeze	4(9.1%)
Cyanosis	3(6.8%)

Chest x-ray findings showed herniation of bowel loops to left hemithorax with mediastinal shift to right in 22 patients (50%), air trapping in right upper lobe5(11.4%) there was, multi cystic area in right hemithorax in 5(11.4%), elevated right dome of diaphragm in 3(6.8%), homogenous opacity in right hemithorax in 4(9.1%), segmental collapse in 2(4.5%) and left lung collapse in 1(2.3%) patient. Computed tomography findings are shown in table 2.

Table 2: Computed tomography of chest findings. N=44

Computed tomography of chest findings	Number (percentage)
Gut herniation in left hemithorax	24(54.5%)
Right lung hyperinflation with trans mediastinal shift	5(11.4%)
Multifocal cystic areas with collapse of right lung	5(11.4%)
Eventration of diaphragm	4(9.1%)
Cavity lesion in right hemithorax	2(4.5%)
Left bronchogenic cyst	1(2.3%)
Left lung hypoplasia	1(2.3%)
Lung agenesis	1(2.3%)
Cyst in lung	1(2.3%)

Regarding diagnosis of children, the most common was congenital diaphragmatic hernia in 24(54.5%), congenital cystic adenomatoid malformation 6(13.6%), congenital lobar emphysema 5(11.4%), eventration of diaphragm 4(9.1%), bronchogenic cyst 2(4.5%), lung agenesis 2(4.5%) and lung cyst 1(2.3%).

Surgery was performed in 33 patients (75%) and not done in 11(25%) out of which in 4(9.1%) cases of eventration of diaphragm were kept on follow up and surgery was not indicated in 2 (4.5%) patients of lung hypoplasia/lung agenesis Three (6.9%) patients with congenital diaphragmatic hernia and 1(2.3%) patient with congenital cystic adenomatoid malformation refused surgery and left against medical advice, one (2.3%) patient of congenital diaphragmatic hernia expired before surgery.

Cardiac anomalies were seen in 3(6.9%) patients, including pulmonary stenosis in 1(2.3%), pulmonary artery hypertension in 1(2.3%) and large left ventricle with large ventricle outflow obstruction and atrial septal defect with pulmonary artery hypertension in 1(2.3%).

Overall, 37 patients (84.1%) survived out of which surgery was performed in 31(93.9%) with post operative survival of 95.4%. Three (6.8%) patients expired, out of which 2(4.6%) had CDH and 1(2.3%) had congenital cystic adenomatoid malformation.

DISCUSSION

Most common diagnosis for congenital thoracic malformations beyond neonatal age was CDH followed by CCAM, congenital lobar emphysema, eventration of diaphragm, bronchogenic cyst and lung cyst. Bentur et al studied congenital thoracic malformations on prenatal ultrasound and found most common finding as congenital pleural effusion.[4] We did not find any case of congenital pleural effusion as congenital pleural effusion usually present in early neonatal period.[4] CDH was the second most common congenital thoracic malformation in their study. Other studies have also documented CDH as most common anomaly in congenital thoracic malformation.[14]

Second most common anomaly we found was CCAM, same finding has been reported by Dincel et al where he found CCAM among the most common lung malformations.[10]Other common anomaly we found were congenital lobar emphysema, pulmonary sequestrations, CPAM and bronchogenic cysts. Similar findings have been reported by other studies.[15,16]

Most common presentation in our patients was respiratory distress followed by cough, feeding difficulty and failure to thrive. This is in concordance with other studies that reported respiratory difficulty as predominant presentation. This is obvious because of mass effect of congenital lung lesion and in case of CDH due to mass effect by herniated bowel loops and may be associated with pulmonary hypoplasia and subsequent development of pulmonary artery hypertension.[17]

Most common computed tomography finding was Gut herniation in left hemithorax which is co related with left sided diaphragmatic hernia. This has been reported that 95% of CDH are Boc dalek hernia , of which 85% occurring on left side of diaphragm.[18] Multi focal cystic area with collapse was also common in our patients which is related to CCAM, reported as the second most common congenital thoracic malformation.[4] We also observed right lung hyperinflation with trans mediastinal shift consistent with congenital lobar emphysema and elevation of diaphragm consistent with eventration of diaphragm which is rare type of diaphragmatic malformation.[18]

Cardiac anomalies were found in 3(6.8%) out of which two had CDH and one has congenital lobar emphysema. Anne et al also showed CDH to be associated with congenital heart disease.[11]

It is recommended that all symptomatic lung malformations should be surgically treated. Regarding treatment of asymptomatic congenital lung lesion there exists controversy for treatment strategy and timing of treatment as well.[4] Present study included all symptomatic patients however, surgery was performed in 33(75%) patients, among rest of the 11 patients, 4 patients (9.1%) refused surgery, 4(9.1%) patients were kept on follow-up observation while 2(4.5%) with lung hypoplasia and lung agenesis did not need surgery and 1(2.3%) expired before surgery.

Studies report a survival rate of 63% in patients with CDH.[19] We have found survival of 93.2% which is high because we only took patients beyond neonatal age. Severe or large congenital defects usually present in early neonatal period while mild to moderate defects can be missed in antenatal or early post-natal period. Such cases then present in late childhood period and are usually not associated with severe pulmonary hypoplasia or pulmonary artery hypertension. It has been previously recorded that early presentation with CDH is associated with poor outcome as compare to late presentation.[20]

Limitations: The present study was conducted in medical units of NICH only hence patients who were directly referred to surgical units might have been missed.

CONCLUSION

Most common congenital thoracic malformation were CDH and cystic adenomatoid malformation. Patients presenting beyond neonatal period showed a good outcome in terms of postoperative survival rate.

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