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Late-presenting congenital diaphragmatic hernia

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ABSTRACT

Background: This study was undertaken to highlight the clinical profile, misdiagnosis, surgical treatment,and prognosis of late-presenting congenital diaphragmatic hernia (CDH) cases in a tertiary level hospital. **Patients and Methods:** This retrospective study included all the babies and children >1 month of age with CDH who were admitted in our Hospital (Sher-i-Kashmir Institute of Medical Sciences, Srinagar, Kashmir, India) during the period between January 2008 and December 2013. Babies with age <1 month were excluded from the study. Data regarding clinical profile, operative records, and follow-up was reviewed and analysed statistically. **Results:** A total of 20 patients were included in this study. The clinical picture ranged from respiratory distress (13 patients) to non-specific gastrointestinal complaints (5 patients). In two patients, CDH was misdiagnosed as pneumothorax and had got chest tube inserted in other hospitals before referral to this tertiary care centre. In 14 patients chest, X-ray revealed the diagnosis of CDH and in remaining five patients (including the two patients with misdiagnosis) further investigations were undertaken to establish the diagnosis. Age ranged from 45 days to 17 years with an average age of 58.9 months. There were 12 male and 8 female patients. In all the 20 patients, surgical procedures were undertaken with the retrieval of herniated contents from the thoracic cavity and repair of the diaphragmatic defect. There was no mortality in our series. All the 20 patients were followed-up for a period ranging from 6 months to 5 years (median 3.1 years). **Conclusions:** Late-presenting CDH can have diverse clinical presentation. Late diagnosis and misdiagnosis can result in significant morbidity and potential mortality if these cases are not managed properly at an appropriate stage. Outcome is favourable if these patients are expeditiously identified and surgically repaired.

Key words: Congenital diaphragmatic hernia, late presentation, misdiagnosis

INTRODUCTION

Late-presenting congenital diaphragmatic hernia (CDH) is much less common and the majority of cases present with nonspecific respiratory and gastrointestinal (GI) symptoms in childhood or early adult life. Presentation beyond the neonatal period varies greatly leading to clinical and radiological misdiagnosis. The prognosis of late-presenting CDH is usually favourable than the CDH presenting in the neonatal period.[1] This study aims to characterize the clinical profile, surgical treatment and prognosis of late-presenting CDH by retrospective review of our experience.

PATIENTS AND METHODS

This study was a retrospective study between January 2008 and December 2013 in a single institution. Records of patients of CDH beyond 1 month to <17 years of age were reviewed retrospectively. Data entered included age, gender, clinical features, radiological findings, and presence of associated anomalies. Furthermore, the operative procedures performed, operative findings, outcome of treatment, and follow-up were recorded in detail.

RESULTS

Out of a total of 100 CDH patients admitted and treated during the study period 20 patients that presented after the neonatal period were included in the study. The median age at presentation was 14 months (range from 1½ months to 17 years). There

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were 12 male and 8 female patients. Thirteen patients presented predominantly with respiratory symptoms including respiratory distress, tachypnea , cyanosis, cough, wheeze and fever. Five patients presented with nonspecific GI symptoms ranging from pain abdomen, vomiting, anorexia and constipation. Two patients were misdiagnosed as pneumothorax in whom intercostal chest tubes were inserted in peripheral health facilities. Abdominal and chest X-ray later in the course of treatment established the diagnosis of CDH. Among these two patients, one patient had a chest tube inserted in stomach located in the thoracic cavity and in other patient chest tube had pierced the colon in thorax. In the first case, the stomach perforation was closed and diaphragmatic defect was repaired and in the second patient colostomy was performed along with the closure of diaphragmatic defect. In 15 patients, only X-ray chest was required for confirming the diagnosis of CDH [Figures 1 and 2]. In five patients, CECT was used to establish the diagnosis. In the rest of the two patients diagnosis was made subsequent to insertion of the chest tubes in the thoracic cavity as CDH was misdiagnosed in these two instances.

Out of 20 patients, four patients required ventilatory support before surgical intervention. In 18 patients, CDH was on the left side. Size of the defect ranged from 2 to 10 cm [Figure 3]. On the left side colon and small bowel was present in all cases. Stomach and spleen were found in 10 and 13 patients, respectively. Malrotation of gut was found in five patients with left sided CDH. Two patients had right sided CDH containing liver, large, and small bowel. The operative approach in the right side defect was abdominal. All the defects were repaired primarily except in one patient where polypropylene mesh graft was used [Figures 4 and 5]. In 15 patients, postoperative course was uneventful. Three patients developed pneumonia in the postoperative period. Out of these two patients required ventilatory support for 3 weeks. In two patients pneumothorax developed on the opposite side post-operatively, which was diagnosed promptly

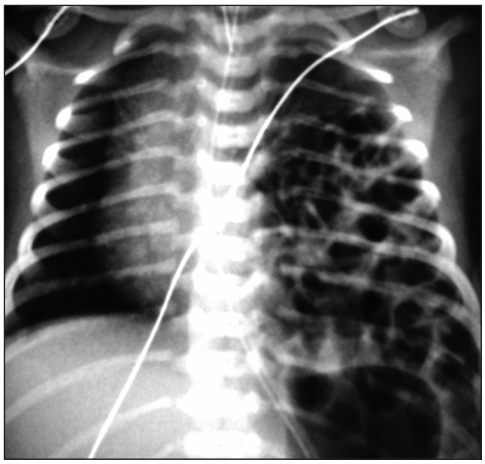


Figure 1: X-ray chest; showing left sided congenital diaphragmatic hernia

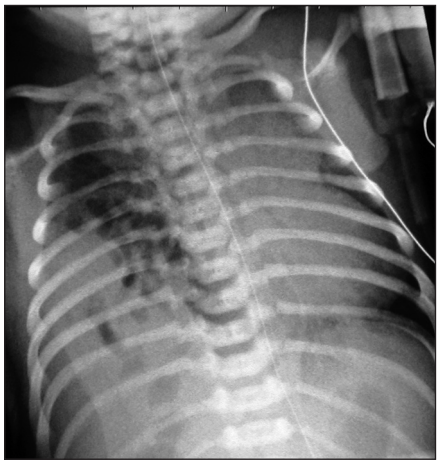


Figure 2: X-ray chest; showing right sided congenital diaphragmatic hernia

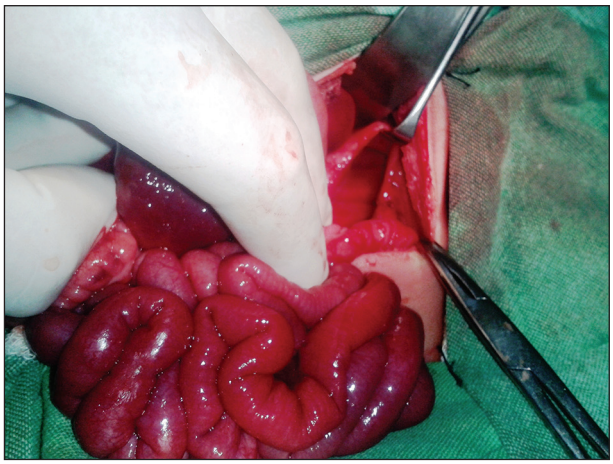


Figure 3: Intra-operative picture; a large left side diaphragmatic defect

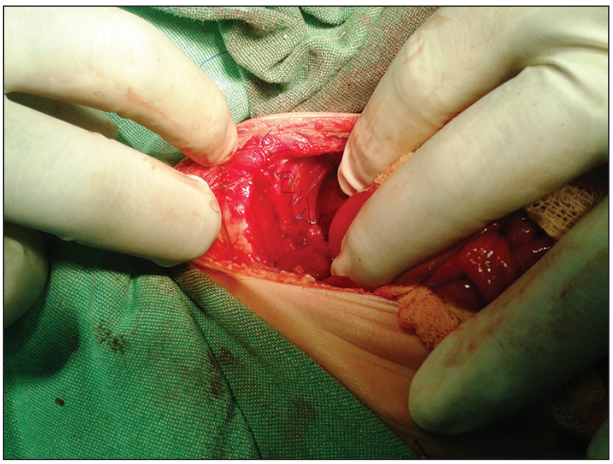


Figure 4: Intra-operative picture showing primary repair of the defect

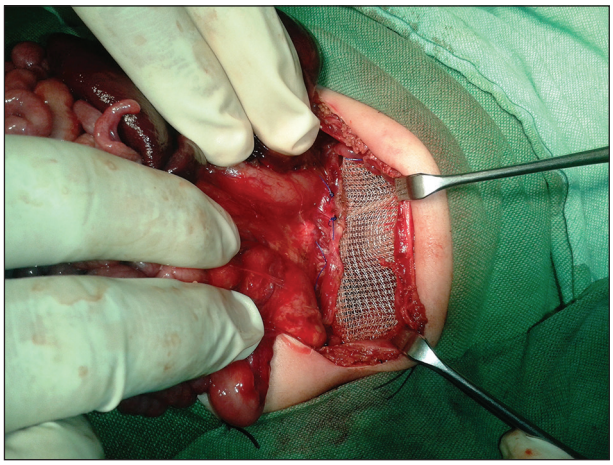


Figure 5: Intra-operative picture; polypropylene mesh used to cover a large defect

and managed by chest tube drainage. Average hospital stay was 9.58 days. Six (15%) patients had associated anomalies. There was no mortality in our series [Table 1].

DISCUSSION

The late presentation of CDH was considered as rare, but an increasing number of publications report that up to a quarter of patients are diagnosed after the neonatal period and are usually associated with a better outcome.[2,3] In our study, the incidence of the late-presenting CDH was 20% and different authors have reported incidence varying from 5% to 25%.[3-5] It is speculated that the increasing trend in incidence may be due to increasing use of imaging in asymptomatic children.

Patients present with a wide variety of respiratory and GI symptoms. At least a quarter of patients are asymptomatic and are often diagnosed incidentally on X-ray chest.[6,7] In previous reports, the most common presenting symptoms were respiratory followed by GI.[8] In our series, respiratory symptoms were the most common presenting symptoms. Kirkland characterised the symptoms of late presentation as grumbling, remittent and misleading and thought that the free movement of abdominal viscera between pleura and peritoneal cavities is responsible for protean characteristic symptoms.[9] Misdiagnosis is often reported in the form of pneumonia and pneumothorax. Chest tubes were inserted in two of our patients, which later were confirmed to be in the colon in one patient and in the stomach in the other patient. Burman *et al*. in their study have reported 4 of 26 (15%) patients diagnosed with tension pneumothorax. Such incorrect diagnosis may be associated with severe morbidity and possible mortality.

Diagnosis is frequently made by X-ray chest, but it is not diagnostic in every instance and it may mimic lower lobe pneumonia, diaphragmatic eventration, pneumothorax, pleural effusion and congenital cystic adenomatoid malformation thereby leading to misdiagnosis. Misdiagnosis and nonspecific presentation often lead to delay in the diagnosis. Such patients need further investigations like computed tomography or contrast study of upper GI tract to arrive at a correct diagnosis. Three patients in our study required such additional investigations for correct diagnosis.

In 19 patients surgical repair was accomplished successfully using Vicryl suture material. Only one patient required polypropylene mesh graft because of a very large defect. The most common organ within the thorax was large bowel followed by small bowel, stomach and spleen [Table 1] which is consistent with the findings of other authors.[10] Five patients had associated malrotation of gut. All patients are doing well on follow-up.

The reported mortality rate in latepresentation of CDH ranges from 8% to 18%, r.[3,11] In our series, there was no mortality. The need for ventilatory support during post-operative period was far less than neonatal CDH. The better prognosis in late-presenting cases of CDH and relatively lesser requirement of post-operative ventilation as demonstrated in our series may be due to a lesser degree of pulmonary hypoplasia in these cases. However, an early and correct diagnosis is the main challenge in such cases. The low mortality rate in our study is probably due to a lower rate of misdiagnosis, lesser incidence of pulmonary hypoplasia, improved surgical technique and appropriate intensive care.

CONCLUSIONS

We conclude that late-presenting CDH can have a wide spectrum of clinical presentation and a high index of suspicion is important for timely and correct diagnosis. It should be suspected in any child presenting with recurrent respiratory symptoms, indeterminate chest X-ray findings and vague GI symptoms. The prognosis is relatively better due to lesser frequency of pulmonary hypoplasia in late-presenting cases of CDH.

Table 1: Clinical characteristics and operative findings of late-presenting CDH

Age/sex (m = month, yr = year)	Side	Clinical picture	Associated malrotation (none/yes)	Herniated contents	Investigations	Site and size (in cm) of the defect	Post-operative course	Hospital stay (days)
1.5 months/male	Left	Respiratory distress	None	Large and small bowel, spleen	CXR	4×4	Uneventful	10
4 months/male	Left	Respiratory distress	None	Large and small bowel, spleen	CXR	5×3	Uneventful	9
1 year/female	Left	Respiratory distress+GI symptoms	None	Large and small bowel, spleen	CXR	4×4	Pneumonia (1 week treatment)	16
3 years/female	Left	Vomiting, fever	None	Large and small bowel, stomach	CXR	3.5×3	Uneventful	7
4 years/male	Left	Respiratory distress	None	Large and small bowel, spleen	CXR+CECT	5×5	Colostomy closed at 6 weeks	22
3 years/male	Left	Abdomen pain, vomiting	Yes	Large and small bowel, stomach	CXR	6×4	Uneventful	11
7 years/male	Left	Incidental finding on X-ray chest for fever	None	Large and small bowel, stomach	CXR	5×5	Post-operative ventilation (1 week) and pneumonia treated	17
2 years/female	Left	Vomiting	None	Large and small bowel, spleen	CXR	5×3	Uneventful	8
6 months/male	Left	Respiratory distress	None	Large and small bowel, stomach	CXR+CECT	5×2	Uneventful	7
10 years/male	Left	After ICT for misdiagnosis of pneumothorax	Yes	Small bowel and stomach	CXR	5×3	Uneventful	9
1.9 years/female	Left	Tachypnoea, cyanosis	None	Large and small bowel, spleen	CXR	3×3	Post-operative ventilation	13
5 years/male	Right	Constipation, pain abdomen	None	Large intestine	CXR+CECT	4×4	Uneventful	6
2.5 years/female	Left	Incidental finding	None	Large and small bowel	CXR	5×2	Uneventful	5
8.5 years/male	Left	Respiratory distress	Yes	Large and small bowel, stomach	CXR+dye study	5×5	Post-operative ventilation (1 week)	11
2.5 years/male	Left	Respiratory distress	Yes	Large and small bowel, spleen	CXR+CECT	6×2	Uneventful	8
4.5 years/female	Left	Constipation	Yes	Large and small bowel, spleen	CXR	4×3	Uneventful	5
2.5 years/male	Left	Fever, vomiting	Yes	Large and small bowel, spleen	CXR+dye study	7×4	Post-operative ventilation	10
6 years/female	Right	Respiratory distress, GI symptoms	None	Large and small bowel, spleen	CXR	PL, 4×3	Uneventful	7
5 years/female	Left	Respiratory distress	None	Large and small bowel, spleen	CXR	5×2	Uneventful	8
9 male/male	Left	Fever, vomiting	None	Large and small bowel, spleen	CXR	PL, 3×5.5	Uneventful	8

CXR: Chest X-ray; CECT: Contrast enhanced computerised tomography; CDH: Congenital diaphragmatic hernia; GI: Gastrointestinal; ICT: Intensive care treatment

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