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Original Research Article

Congenital Diaphragmatic Hernia beyond the Neonatal Period: Diagnostic Challenges and Management

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Abstract

Background: Infants and Children with Congenital Diaphragmatic Hernia (CDH) presenting after the neonatal period have varied presentations from incidental radiological finding to severe complications like intestinal obstruction and gastric volvulus.

Methods: A retrospective review of the medical records between January 2014 and December 2024 of the infants and children admitted with congenital diaphragmatic hernia presenting after the neonatal period at our institution was performed.

Results: A total of 15 patients of CDH presented after the neonatal period. Respiratory distress was the commonest presentation. Intestinal obstruction was another common presentation with few cases of volvulus of the stomach. There were three deaths (20%) and 12 survivors (80%).

Conclusion: The diagnosis of CDH after neonatal period is challenging with varied presentations including intestinal obstruction and Gastric volvulus. Prompt surgical management with appropriate perioperative critical care are necessary to ensure survival.

Keywords: Congenital Diaphragmatic Hernia, Intestinal Obstruction, Gastric Volvulus.

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Introduction

Antenatal diagnosis and neonatal presentation with respiratory symptoms are the common presentation of Congenital Diaphragmatic Hernia. However, a small subset of CDH can present later in the infancy or in the early childhood with a wide variation of symptoms. The incidence of this presentation after neonatal period is reported to be between 5 – 45% [1,2]. The presentation of this subset of CDH patients is very different from the newborn presentation and survival depends on timely surgical intervention.

Materials and Methods:

The medical records of children diagnosed with CDH who were treated at the Institute of Child Health and Hospital for Children, Madras Medical College, and Chennai were retrospectively reviewed. The study period was for 10 years from January 2014 to December 2024. All the infants and children with CDH confirmed during surgery were included in the study. The neonatal CDH patients and children with eventration were excluded from the study. The variables analysed include age, sex, symptom, surgical procedure, and overall survival. The emphasis was on presenting signs and symptoms of the patient. Patients underwent Chest X-ray and CT scan of the Chest. An echocardiogram was done to rule out pulmonary hypertension. The need for perioperative mechanical ventilation was based on the clinical condition of the patient.

Results

A total 15 patients who were diagnosed with CDH after 30 days of age were included in the study. There were nine male children and six female children. Age wise, eight children were infants, five were less than five years of age and two were above five years of age. Twelve patients had left-sided defect and three children had right CDH. The clinical and the operative details are summarised in the Table 1.

A total of five children presented with severe respiratory distress and required mechanical ventilation. The respiratory distress was less severe in the six patients. F our patients had upper respiratory tract symptoms (URI) without distress

and were diagnosed with CDH in the screening chest X-ray.

Three patients presented with intestinal obstruction. One infant had a knuckle of the transverse colon was found trapped in a small 2 x 1 cm circular defect in the left posterolateral diaphragm. The child underwent resection of the ischemic colon and anastomosis and subsequent closure of the defect. Second child had presented with acute small bowel obstruction due to constriction at the level of defect (Figure 1). After successful reduction, bowel perfusion improved. Third child presented with colonic obstruction and had perforated appendicitis and pus in right thorax during exploration.

Three patients presented with gastric volvulus. One of them had intraabdominal gastric volvulus and presented with abdominal pain and severe retching. The X-ray chest and abdomen was reported as left eventration with prominent stomach. During surgery, a 3 x 3 cm left diaphragmatic defect with herniated small bowel and volvulus of the stomach was found. A CDH repair and an anterior gastropexy was done.

The other two patients had intra-thoracic gastric volvulus. One child presented with gangrene of the intra-thoracic stomach with severe shock. The child died due to a cardiac arrest on table during the emergency surgery. Another patient with intra-thoracic stomach and gastric volvulus with severe respiratory distress on ventilator was wrongly diagnosed as left pyo-pneumothorax based on the chest X-ray and an intercostal drain was inserted. Later the CT Chest was performed and a diagnosis of left CDH was made and the child was operated. The child required prolonged post-operative ventilation and underwent re-operation for burst abdomen and finally died of ventilator associated pneumonia.

The infants and children who were stable were evaluated with an Echocardiogram. None of them had pulmonary hypertension. One child had small ASD. Three children underwent CT chest to confirm the diagnosis. Eventration was the most common differential diagnosis and the diagnosis was confirmed only during the surgery.

All the children underwent laparotomy and repair of the diaphragmatic defect. The defect was small less than 3 cm in six patients and larger than 3 cm in the remaining patients. All of them had a tension-free anatomical repair of the diaphragm.

Perioperative mechanical ventilation was required in five patients who presented with severe respiratory distress and intra-thoracic gastric volvulus. None of the remaining patients required ventilatory support. An intercostal drain was placed in all patients and was typically removed after 48 to 72 hours.

The two children with intra-thoracic gastric volvulus and one child with prolonged postoperative ventilation died in this study. The remaining 12 patients survived. All the surviving children are on regular follow-up and there was no documented recurrence in any of the patients.

Table 1: Clinical features, complications and survival of Children with CDH

S. No.	Age/Sex	Symptom	Surgery	Mech vent	Echo	Complications	Survival
1.	3/12, M	Intestinal obstruction	CDH repair/ colon Resection	No	N	Nil	Yes
2.	4/m	Gastric volvulus	Left CDH repair	No	N	Prolonged NG aspirate	Yes
3.	7/m	Intrathoracic Gastric volvulus	Left CDH repair	yes	Not done	Gangrene stomach, shock	No
4.	8/12/M	Incidental/URI	Left CDH repair	No	N	Nil	Yes
5.	6/M	Intrathoracic Gastric volvulus	Left CDH repair	yes	N	Prolonged post op ventilation, burst abdomen.	No
6.	11/12/f	Incidental /URI	Left CDH repair	No	N	Nil	Yes
7.	1 ½ m	Pneumonia	Left CDH repair	No	N	Nil	Yes
8.	7/12/f	Bronchiolitis	Left CDH repair	No	N	Nil	Yes
9.	1 3/12 m	Recurrent URI	Right CDH repair	No	N	Nil	Yes
10.	2 ½ m	Pneumonia	Left CDH repair	No	N	Nil	Yes
11.	4 / m	Failure to thrive /URI	Left CDH repair	No	N	Nil	Yes
12	4/12 F	Intestinal obstruction	Right CDH repair	Yes	Y	Postoperative ventilation	Yes
13	6/12 F	Intestinal obstruction	Right CDH repair	Yes	Y	Postoperative Ventilation	Yes.
14	5/F	Retching	Left CDH repair	No	Y	No	Yes
15	2/12 F	Respiratory distress	Left CDH repair	Yes	Y	Pneumonia	No

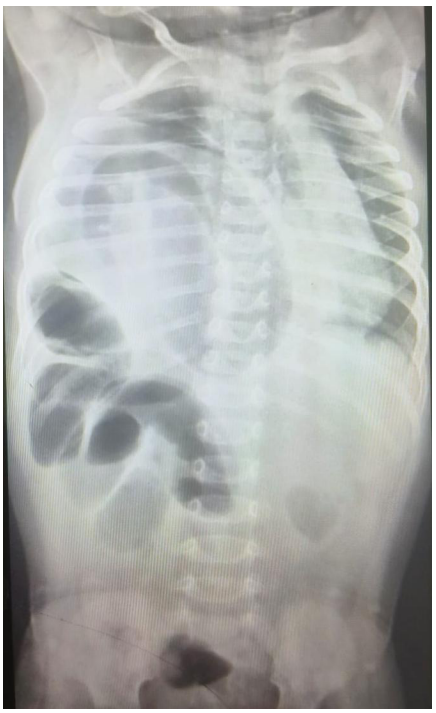

Figure 1: Right CDH with intestinal obstruction

Discussion

Children with CDH presenting after neonatal period had both respiratory and gastrointestinal symptoms. Respiratory distress remained the most common presentation.

However, cause of respiratory distress was usually an underlying pneumonia or rarely due to gastric volvulus rather than the pulmonary hypoplasia and pulmonary hypertension which are common in the neonatal CDH patients.

The cause of death in our study was due to the intra-thoracic gastric volvulus. The presentation of gastric volvulus can be acute in an apparently normal child [3]. Can be confusing because the dilated intrathoracic stomach in Chest X-ray typically mimics a lung cyst or a pyo-pneumothorax [4] necessitating a CT scan of the Chest. CT chest is the gold standard test to identify delayed CDH and especially an intrathoracic gastric volvulus. Gastric volvulus that presents acutely is a surgical emergency and immediate surgery is indicated if the diagnosis is confirmed with a chest X-ray. Early identification and timely intervention will prevent morbidity and mortality. Apart from this small subset of the patients who presented acutely with gastric volvulus, the remaining patients had excellent prognosis. Some Children presented with intestinal obstruction and X-abdomen revealed CDH. The defects are typically small causing constriction of the bowel causing intestinal obstruction. Immediate surgery without delay is recommended for these patients. Open diaphragmatic hernia repair was performed in all cases although laparoscopic or thoracoscopic repair is feasible and reported [5].

To conclude, after the neonatal period, CDH usually presents with either respiratory distress or symptoms of intestinal obstruction and gastric volvulus. These children require immediate surgery after stabilization. The survival rate depends on early intervention without delay as bowel gangrene results in major morbidity.

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