ORIGINAL ARTICLE

Congenital Anomalies of the Diaphragm in Children

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Abstract

Twenty children with various diaphragmatic anomalies, managed over a period of 3 years, are presented. These included congenital diaphragmatic hernia (CDH 10), congenital eventration of diaphragm (CDE 6) and hiatus hernia with gastro-esophageal reflux (HH-GER 4). Clinical presentation of these children was : respiratory distress (7), recurrent respiratory tract infection with failure to thrive (11) and gastric volvulus (3). Two patients of gastric volvulus had acute surgical abdomen. The patients in respiratory distress group were all newborns with CDH. Whereas, those who had gastric volvulus were CDE 2 and HH-GER 1. All children were operated through left subcostal transabdominal approach, except those with HH-GER who were explored through upper midline incision. Repair of diaphragmatic defect/plication was done in patients with CDH/CDE, whereas Nissen fundoplication (Loose wrap) was done in patients of HH-GER. Three newborns with CDH and one child with CDE and gastric volvulus died. All other 16 surviving children are doing well after 4 months to 3 years of follow up.

Key words

Diaphragmatic hernia, Eventration, Gastro-esophageal Reflux, Hiatus Hernia, Volvulus.

Introduction

Congenital anomalies of the diaphragm and its various hiatuses are not uncommon in children. These include posteriolateral hernia of Bochdalek, the substernal hernia of Morgagni and various degrees of eventration or paralysis of the diaphragm. In some cases, part of the stomach may herniate through a lax esophageal hiatus, causing a hiatus hernia which may or may not be associated with GER. A Bochdalek hernia is one of the most important and common surgically correctable cause of respiratory distress in the newborn. Despite advances in the diagnosis and treatment of birth defects, there remains a considerable mortality in babies born with this anomaly. In other diaphragmatic defects, the surgical mortality is low unless complicated with other severe congenital anomalies. This study was done with an objective of analysing the clinical spectrum and outcome of various anomalies seen at our center.

Patients and Methods

Twenty infants and children having various defects of diaphragm managed over a period of 3 years (Jan. '1994 to Dec.'1997) are reported. There were 16 males

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and 4 females in this study. The age and etiology of various congenital anomalies of the diaphragm (CAD) are shown in table 1. The clinical presentation was closely related to the age and the cause of the diaphragmatic defect. All newborns presented with respiratory distress (RD) due to CDH. Patients of CDE and those of CDH, presenting later than neonatal period and those with hiatus hernia, had recurrent respiratory tract infections (RTI), failure to thrive, and symptoms in all these patients could be traced to early infancy. A combination of symptoms were present in significant number of patients as shown in table 2. Two patients of CDE and 1 patient with HH had gastric volvulus (GV). Table 2 depicts the clinical findings in various patients of CAD in this study. In one patient of CDE, the defect was recurrent whereas in another patient HH-GER occurred 6 months after repair of CDE. Diagnostic investigations in these children were : abdomen and chest x-ray (AXR-CXR), barium esophagogram and arterial blood gas analysis where ever possible. All patients except one of GV, who died in the preoperative period, were operated through left subcostal trans-abdominal/ midline incision. Repair of the diaphragmatic defect/ plication of CDE was done with or without intercostal drainage (ICD) in patients of CDH and CDE. Patients of HH-GER were treated by Nissen's fundoplication (Loose wrap). In two patients (CDE 1, HH-GER 1) who had gastric volvulus, anterior gastropexy was done in addition to fundoplication. Various surgical procedures done for CAD are shown in table 3.

Results

The diagnosis of CAD was established in all patients with clinical data and supplementary radiological workup. All cases of CDH and CDE in our study were on the left side. In newborns, AXR-CXR were sufficient to confirm the diagnosis of CDH, whereas contr studies were done on two occasions in childr presenting later on. The presence of HH-GER w diagnosed on barium studies. One of these infants ha large air fluid level on CXR due to intra-thorac stomach. Barium meal in this patient revealed a part volvulus of the stomach in the posterior mediastinum In one patient of CDE, the GV was confirmed on bariu study. Operative findings in cases of CDH revealed defect of 4 to 6 cms. in the postero-lateral aspect of t diaphragm with varying lengths of small and large bowe stomach, left lobe of liver in all cases and in 3 case spleen was also seen in the chest cavity. The repair o the defect was possible without any prosthetic graft a difficulty in closing the abdominal cavity. In 2 patient of CDH, the posterior leaf of the diaphragm was missin and Gerota's fascia was used to reinforce the posterio limit of the diaphragmatic defect. Left lung was grossl hypoplastic in 2 cases with just a tongue of tissue at th thoracic apex. All patients of CDE could be plicated easily except in 1 who had recurrent eventratio following surgery 3 years back. Similarly one patien who had HH-GER after repair of CDH, presented difficulty due to adhesions around the left side of cardio-esophageal junction. Two newborns with CDH, who had severe hypoplasia of the lung, died of respiratory failure in the post-operative period. Another newborn died of septicemia. One patient of GV died pre-operatively possibly because of gastric necrosis leading to hypovolemic shock. Interestingly no major congenital anomalies of any other system were found in these babies. All the surviving patients of CDH/CDE are free of any recurrence, whereas, those of HH-GER are free of their symptoms and are thriving well after four months to three years of follow up.

Discussion

CDH is one of the commonest causes of respiratory distress in neonates. The incidence of this anomaly varies from 1 in 2000 to 5000 live births with a greater incidence in still births and abortions (1). Embryologically, the most common type of the CDH (Posterio-lateral hernia of Bochdalek) occurs becuase of failure of pleuro-peritoneal canal to close. The exact etiology of congenital diaphragmatic hernia is not known, however, vitamin A deficiency and use of drugs like Phenmetrazine, Quinine, Thalidomide, Bendectine and Nitrophen have all been implicated for this (1). Posterio-lateral defects account for 75 to 85% of CDH and are more common on left side. Whereas left sided defects are more common and present in neonates, the right sided defects present late in older children. Bilateral defects are rare (1%) and are usually fatal (2). Excluding malrotation and patent ductus arteriosus, the incidence of the associated anomalies is between 10 to 20%, and it may rise to 95% in still born (3) Complex cardiac defects, such as, Cantrall's pentology may be present. Diagnosis of CDH is not difficult. A careful examination of the baby will reveal the features of respiratory distress, scaphoid abdomen and increased antero-posterior diameter of the chest. A plain AXR/CXR is classical with loops of intestines in the hemithorax, absence of hemidiaphragm on the affected side with paucity of abdominal gas shadows and mediastinal shift. With increasing use of prenatal ultrasound, the diagnosis of CDH may be detected in utero (4). Once the diagnosis of CDH is made, the treatment is surgical after resuscitation and stabilization of the baby. It has been seen that improvement in pre-operative arterial blood gas (pH>7.2, $PaCO_2$<50 mm of Hg and PaO_2>100 mm Hg) and alveolar-arterial oxygen difference (A-aDO_2) of less than 500 mm of Hg are associated with better survival (5,6). The use of intercostal drainage (ICD) after repair of CDH is debatable and some workers even suggest bilateral ICD to avoid the chance of contralateral pneumothorax which is reported in 7 to 20% of the patients with considerable mortality (7). Post-operatively, it is mandatory to carefully monitor the patients of CDH to avoid the various complications (pneumothorax, respiratory failure, pulmonary hypertension and persistent fetal circulation) and early mortality. A ventilatory support, use of vaso-dilator drugs and extra-corporeal membrane oxygenator (ECMO) may be required to reverse pulmonary hypertension and persistent fetal circulation (PFC). Inspite of these advancements in management, CDH still remains a potentially lethal congenital anomaly. The mortality rates for babies who are live born and become symptomatic within 6 hours of life remains at 50%, while those who present after 24 hours of birth have a 100% chance of survival (8). Although, survival rates may be improved by 10-15% through high frequency ventilation and ECMO in early presentation group, some of these babies, because of severe pulmonary dysplasia are unsalvageable even with these modalities (9, 10). Lung transplant may prove to be a potential treatment of such babies using ECMO as a bridge to transplant (11). Fetal surgery, which is still in experimental and infancy stage, may provide the much needed solution (12).

The other anomaly affecting the diaphragm is eventration. It has been defined as "an abnormally high or elevated position of one leaf of the diaphragm as a result of *paralysis, aplasia or atrophy* of various degrees of the muscle fibres" (13). CDE is a developmental abnormality due to partial / no muscularization of septum transversum from the myotomes of the adjacent body

wall (14, 15). It is a rare disorder with 3 cases affecting every 10,000 neonates (16). Male and left sided predominance is noted. Associated anomalies may be present in a significant number of cases, the commonest being the cardiac and chromosomal, which have a strong bearing on the outcome of treatment. All symptomatic patients require surgery through laparotomy/thoracotomy approach.

Hiatus hernia in a child may be due to congenital or acquired causes. The congenital type is rare, whereas, the acquired one may occur in 5.9 to 16.8% of cases, mainly after Nissen's fundoplication for GER. The cause may be disruption of the repaired crura, or constant pressure of gastric fundus on diaphragm. An added factor may be peri-esophageal dissection (17-19). Congenitally the hiatal structures including diaphragm, gastro-esophageal ligaments along with gastrocolic and gastrosplenic attachments may have varying degree of weakness potentiated by contributory factors like : high intra-gastric pressure in the presence of pyloric narrowing, gastric redundancy and the effects of negative intra-thoracic pressure (19). Hiatus hernia may be asymptomatic which suggests the possibility of spontaneous intermittent reduction. A careful history and high degree of clinical suspicion is needed in patients with recurrent RTI, regurgitation of feeds and failure to thrive to rule out HH. A chest X-ray and upper GI series are essential to reach at the diagnosis. Endoscopy may be needed to supplement radiological investigations (18). The current surgical treatment of HH in infancy and childhood is laparotomy, proper inspection of upper GI tract, reduction of hernial contents, careful dissection of gastro-esophageal junction, narrowing of crura, partial or complete excision of the hernial sac and an anti-reflux procedure. Some authors recommend anterior gastropexy to decrease the possibility of recurrence of HH (20).

In conclusion, we believe that congenital anomalies of diaphragm present a spectrum ranging from diaphragmatic hernia to hiatal defects. Each anomaly has a separate embryological basis. The time of prese and outcome of these anomalies is primarily rel the severity of the defect. Diaphragmatic hernia most common of these anomalies with prognosis d related to the factors like, the age of patient, seve pulmonary dysplasia and associated anomalies. CD HH carry a good prognosis as evidenced by ou results.

Table 1. Age and etiology of CAD (n = 20

Age	Diagnosis			Total N of case
	CDH	CDE	HH-GER	
Newborn	08	–	–	08
Infants	02	–	03	05
1-3 years	–	02	01	03
3-6 years	–	04	–	04
Total :	10	06	04	20

Table 2. Clinical Presentation of CAD (n = 2

Diagnosis	Clinical Presentation			
	RD	RTI	Failure to Thrive	Acute Abdom
CDH	08	02	02	–
CDE	–	06	06	02
HH-GER	–	04	04	–
Total :	08	12	12	02

Table 3. Operative Procedures & Results of CA

(n = 19)**

Diagnosis	Operation	No. of cases	Results
CDH	Repair ± ICD	10	3 Newborns died (Resp. failure=2, Septicemia=1)
CDE	Plication*	05(1)**	All survived
HH-GER	Nissen fundoplication* (Loose wrap)	04	All survived

*Gastropexy was added in 2 patients with gastric volvulus.
**One patient of gastric volvulus died preoperatively.

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