

Case Report:

Congenital diaphragmatic anomalies: Case review

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Abstract:

This series of case reports presents a review of three cases of congenital diaphragmatic anomalies. Two cases are that of congenital diaphragmatic hernia, one, a patient brought to the hospital at 11 months of age with complaints of cough and grunting for one month. Another case aged 44 years presented with complaints of severe abdominal pain and vomiting for 1 day. The third case was a patient diagnosed with diaphragmatic eventration at the age of 26 years who presented with complaints of breathlessness on exertion and a dragging type of pain on the left side of the chest for a period of 1 month. All three cases were admitted and treated in Sri Manakula Vinayagar Medical College and Hospital, Madagadipet, Pondicherry. An effort will be made to compare and give an explanation for the presentation and timing of the three cases in the report.

Keywords: Congenital diaphragmatic hernia

Introduction:

The thoraco- abdominal diaphragm is a musculo-aponeurotic partition between the thoracic and abdominal cavities. Congenital diaphragmatic hernia (CDH) has been described since the 17th century. Its estimated incidence has been reported to be 1 in 2000–5000 live births. Diaphragmatic hernias through the posterolateral foramen of Bochdalek represent the commonest type of congenital diaphragmatic hernia^[1]. Some authors have suggested that if the herniation is present from the time of birth, it is termed “congenital”. If the herniation forms later, it is termed “acquired”^[2]. Late presentation of CDH is uncommon, and accounts for 5-30% of all CDH cases^[1]. The mean age of diagnosis in a case of congenital diaphragmatic hernia is by 372 days^[1]. This emphasizes the rarity of the report as two of the cases present at 44 years and 26 years with delayed

presentation of a diaphragmatic developmental defect.

Case reports:

A male baby, aged 11 months, weighing 7.5 kg, who had been recently started on semi- solid feeds, presented to the surgical outpatient department with complaints of cough and grunting for one month. On examination, the baby was afebrile and conscious. On examination of the chest, bowel sounds were heard on the left side of the chest, breath sounds were reduced on the left side, while the right side of the chest presented normal vesicular breath sounds with laboured breathing. The apex beat was shifted to the right. Per abdomen examination revealed a soft, non- tender abdomen and no organomegaly. Bowel sounds were heard in the abdomen too. On radiological examination, coils of intestine were visualised in the left hemithorax. The baby did not show any signs of respiratory distress and was posted for elective

surgery. Under general anaesthesia, the abdomen was opened via a left subcostal incision and findings noted. Intra-operative findings confirmed the case to be a Bochdalek's hernia. The jejunum, about 10 cm distal to the duodeno- jejunal junction, the ileum and the transverse colon were found to be herniating through a posterior defect in the diaphragm into the left thoracic cavity. The surgery done was- Laparotomy with reduction of contents and anatomical hernia repair. All the contents were gently reduced and placed in the anatomical position. The posterior defect of the diaphragm was repaired by interrupted sutures using 2-0 prolene. The abdomen wound was closed in layers. The baby recovered well and was discharged in 10 days.

The second case was that of a 44 year old male presenting with complaints of severe abdominal pain and vomiting for 1 day. On eliciting the history, the patient revealed that the vomiting was bilious in nature. There was no history of thoracic trauma. On examination of the chest, breath sounds were reduced and bowel sounds heard on the left side. The apex beat was normal in position. The right side of the chest revealed normal vesicular breath sounds. Per abdomen examination revealed guarding. Tenderness was noted in the periumbilical area. Bowel sounds were heard. The patient had mild electrolyte imbalance, after correction of which ultrasound of the abdomen was performed. The ultrasound showed mild hepatomegaly and minimal ascites. The CT scan revealed a mildly displaced spleen along with bowel loops in the left hemithorax. The patient was taken up for emergency surgery- Reduction and repair of rent with Splenectomy. Under general anaesthesia, the abdomen was opened via a midline incision extending from the xiphoid process till two cm below the umbilicus and intraoperative findings were noted. A large defect was found in the

posterior aspect of the diaphragm, through which loops of the small bowel, transverse colon up to the splenic flexure, had herniated into the left thoracic cavity. The loops of the small intestine were covered by a peritoneal bag (Figure 1). The left lung was hypoplastic (Figure 2). The contents were gently retracted and placed in their anatomical position. The herniated contents were normal in size. Splenectomy was performed for want of volume for the retracted contents. Interrupted sutures using 1-0 prolene were put to close the defect in the left posterior part of the diaphragm. Drainage tubes were placed both in the abdomen as well as for inter- costal drainage. The patient recovered well after the surgery and was discharged after 2 weeks.

The third case was that of a 26 year old male patient, presenting with complaints of breathlessness on exertion for a period of 1 month and a dragging type of pain on the left side of the chest. On examination, breath sounds were reduced on the left lower zone of the lung. The apex beat was normal in position. Per abdomen examination revealed a soft, non-tender abdomen, bowel sounds were heard. On radiological investigation, the thoraco abdominal diaphragm on the left side was elevated with the stomach protruding into the lower thorax causing atelectasis of the lower lobe of the left lung. The patient was taken up for emergency surgery- double breasting of the left hemidiaphragm. Abdomen was opened via a left subcostal incision and intra- operative findings recorded. The continuity of the diaphragm was maintained, as such there was no gaping defect. However, a major part of the left dome of the diaphragm was considerably thinned out. The stomach, greater omentum, spleen, splenic flexure and transverse colon were found protruding into the lower part of the thorax. The contents were gently laid in their anatomical position. Double breasting

of the thinned out portion of the left dome was done using 1-0 prolene interrupted sutures. The anterior leaf of the central tendon was approximated with the muscular part. The postoperative period was uneventful and the patient was discharged after 1 week.

Discussion:

The diaphragm divides the thorax from the abdomen and is derived from mainly four sources during embryological development. Initially it is situated rostral to the somites and acts as a mesodermal bridge between the pericardial and the umbilical vesicle cavities. The cranial portion is covered by pericardium and pleura, whereas the ventral part is a sagittal mesentery containing the expanding liver. The septum transversum gives rise to the majority of the diaphragm. It descends from the cervical to thoracic level by the eight week. The other parts of the diaphragm consist of the pleuroperitoneal membranes, dorsal mesentery of oesophagus and muscular ingrowth from the body wall. The pleuroperitoneal canals are closed by the contribution of the adjacent organs, such as the liver and the suprarenal glands. Congenital Diaphragmatic hernia is a displacement of the abdominal organs into the thoracic cavity through a weak area or a distinct defect in the diaphragm [3]. It results from a lack of closing of the pleuroperitoneal cavity by incomplete diaphragmatic development before the intestine returns to the abdomen from the yolk sack between weeks 8 and 10 of gestation [4]. The left-sided Bochdalek hernia occurs in approximately 85% of cases. Left-sided hernias allow herniation of both the small and large bowel and intra-abdominal solid organs into the thoracic cavity. In right-sided hernias (13% of cases), only the liver and a portion of the large bowel tend to herniate. Bilateral hernias are uncommon and usually fatal [2]. The majority of patients with Bochdalek hernia present during

neonatal life and have a poor prognosis, being associated with congenital pulmonary abnormalities, while studies have concluded that the prognosis of late-onset CDH is excellent once the correct diagnosis is made. Presenting symptoms of late onset CDH could be classified as respiratory (upper respiratory tract infection, pneumonia, respiratory distress, cough, wheezing, etc.), gastrointestinal (vomiting, abdominal pain, failure to thrive, constipation, etc.), both or asymptomatic. Gastrointestinal symptoms predominate in left sided hernias whereas respiratory symptoms are more common in right-sided lesions [5].

Eventration of the diaphragm is an abnormal elevation of the dome of diaphragm. It is a condition in which all or part of the diaphragm is largely composed of fibrous tissue with only a few or no interspersed muscle fibers but its continuity is maintained. It can be complete or partial [6].

The significant points noted while reviewing the three cases mentioned in this series of case reports as well as the relevant literature are as follows:

1. In the case of the 11 month old baby with CDH, the baby presented with mild symptoms. Findings were obvious on physical examination. Intra-operatively, while the lungs did not show any abnormality, the intestinal loops were slightly hypoplastic. Given that the baby had been recently started on semi-solid diet, and the functioning state of the intestine, gastrointestinal symptoms were not very evident. One can only assume that in case of further delay in the diagnosis, the baby could have presented with both respiratory as well as gastrointestinal symptoms. There is also a suspicion that the symptoms of the baby might have been obvious earlier had it been the hypoplasia of the lungs rather

than that of the intestine. Since the patient did not show signs of moderate or severe distress, the surgery was done electively.

2. With regards to the second case, the 44 year old patient presented with sudden gastro intestinal symptoms of severe abdominal pain and bilious vomiting leading to the initiation of electrolyte derangement. The intra operative findings of the patient revealed a hypoplastic lung on the left side with normal sized intestinal loops. The patient's body had probably adjusted to the condition and he presented only when complications arose from the herniated loops of intestines. The patient presented with severe distress and could have gone for further dire complications like ischemia or gangrene had it not been diagnosed. This case gives an insight into the statement that diaphragmatic hernias should be surgically corrected by anatomical repair as and when diagnosed, so that morbidity and mortality due to diaphragmatic hernia can be prevented.
3. The third case was a patient with diaphragmatic eventration. The patient was brought with complaints of breathlessness and a dragging pain on the left side of the chest for a period of 1 month. The patient's radiological examination revealed that there was atelectasis of the left lung due to pressure by the protruding organs into the lower left hemithorax. Since he was symptomatic, he was taken up for emergency surgery. As such in cases of diaphragmatic eventration, the continuity of the diaphragm is maintained, and only

in symptomatic cases patients are advised to go for surgery.

Congenital diaphragmatic anomalies including hernia or eventration are an uncommon diagnosis in adults. The knowledge of this anatomic defect presenting among adults is vital for its diagnosis and management, as it should be surgically corrected to prevent complications or to correct them if they have already developed. Because of its varied clinical presentation, it is challenging to make a clear-cut diagnosis of congenital diaphragmatic anomalies. The severity and timing of its clinical presentation almost always depends on the state of the lungs, the herniated contents and how well the body adapts to the condition. A high degree of clinical suspicion, chest radiograph and CT scanning remain the key to a prompt diagnosis in most cases.

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Figure 1: Intra- operative view of the second case, the 44 year old man with late presentation of Bochdalek Hernia, showing the peritoneal bag covering the loops of the small intestine.



Figure 2: Intra- operative view of the second case, the 44 year old man with late presentation of Bochdalek Hernia, showing the hypoplastic left lung through the large diaphragmatic defect



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