RIGHT SIDED CONGENITAL DIAPHRAGMATIC HERNIA-A CASE REPORT WITH A BRIEF REVIEW

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ABSTRACT
Congenital diaphragmatic hernia occurs in 1 in 2000-4000 live births and accounts for 8% of all major congenital anomalies. Congenital diaphragmatic hernia (CDH) is a major surgical emergency in newborns because the key to survival depends on the prompt diagnosis and treatment. We are presenting here one such congenital diaphragmatic hernia. In North Bengal Medical College a fullterm female baby was delivered with respiratory distress, scaphoid abdomen and cyanosis. Immediately chest X-ray and CT scan of thorax and abdomen was done to assess symptomatically and clinically suspected case of congenital diaphragmatic hernia. It was a case of right sided congenital diaphragmatic hernia. The presented case report might help us to remain conscious about such congenital anomaly in a case of respiratory distress and help us to chalk out the immediate measurements accordingly to save lives of those unfortunate newborns.

Key word: Congenital diaphragmatic hernia, Diagnosis and Approach.

INTRODUCTION
The congenital diaphragmatic hernia (CDH) was first described in medical literature in 1679 by Lazarus Riverius who incidentally noted a CDH during a postmortem examination of a 24-year old person. In 1761, Giovanni Battista Morgagni discussed congenital diaphragmatic hernia and other diaphragmatic hernia elaborately. He described the classic anterior diaphragmatic hernia and today it is named Morgagni hernia.

In 1848, Victor Alexander Bochdalek, a professor of anatomy in Prague, described both right and left posterolateral CDH and today it is referred as Bochdalek hernia.[1]

So the congenital diaphragmatic hernia may be of three basic types and those are posterolateral Bockdalek hernia (usually occurring at approximately 6 weeks of gestation), the anterior Morgagni hernia and the hiatal hernia. Most common type of CDH is left-sided Bockdalek hernia (85%). In left sided hernia the large and the small bowel with or without intraabdominal solid organ may be herniated into the thorax. In right-sided hernia (incidence 13%) only the liver and portion of small bowel tend to be herniated into the thorax.[2]

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Mortality in babies with congenital diaphragmatic hernia depends on the severity of the pulmonary hypoplasia and pulmonary hypertension which is associated with a decrease in cross-sectional area of pulmonary vasculature and dysfunction of the surfactant system. Pulmonary hypoplasia along with surfactant dysfunction will lead to small alveolar capillary membrane and the gaseous exchange will become jeopardized. Pulmonary hypoplasia may be complicated by increased muscularization of the intraacinar pulmonary arteries and occasionally in very severe cases left ventricular hypoplasia. So, congenital diaphragmatic hernia is an emergency as the pulmonary capillary blood flow is decreased because of the small cross sectional area of the pulmonary vascular bed and becomes much complicated when it is associated with pulmonary vasoconstriction. Hypoplasia of the lungs is usually associated with long standing and large congenital diaphragmatic hernia and these infants are less likely to survive after birth.[1]

Congenital diaphragmatic hernia may be complicated when it is associated with other anomalies like gastric volvulus, rotational abnormalities and midgut volvulus (incidence rate 30-62%), gastric or intestinal perforations and left ventricular hypoplasia of heart, etc.[3]

Congenital diaphragmatic hernia (CDH) may occur as a nonsyndromic or isolated defect. Only 2% of the CDH cases are estimated to be familial. Familial transmission may be associated with autosomal recessive, autosomal dominant, and X-linked.

Comelia de Lange syndrome is such an autosomal dominant syndrome which is associated with congenital diaphragmatic hernia along with facial anomalies, hirsuitism and developmental delay. Fryns syndrome is an autosomal recessive abnormality where the main defect is the congenital diaphragmatic hernia along with other anomalies like hypoplasia of the distal digits, abnormalities of the brain, heart and genito-urinary system.[1]

No single gene mutation has been identified as producing or contributing to this anomaly and 4% cases are reported to be associated with karyotype abnormalities. CDH may be found in various chromosomal anomalies, including trisomy 13, trisomy 18, and tetrasomy12P mosaicism. In Pallister-Killian syndrome (tetrasomy12P mosaicism) the features are mostly like Fryns syndrome.[1] Chromosome deletions of 1q, 8p and 15q have been reported in association with congenital diaphragmatic hernia, whereas deletions of chromosome 8p and 15q appear to be associated with heart malformations.[3] Congenital diaphragmatic hernia was detected in Brachman-de Lange syndrome in which a NIPBL gene mutation was identified. Brachman-de Lange syndrome the variations were an abnormal peak at the 29th exon in the translation area of the NIPBL gene and a mutation of cytosine to thiamine (nonsense mutation) at the 5524th base.[4]

Here a uncommon right sided (left-sided CDH more common) congenital diaphragmatic hernia (CDH) in a newborn female baby has been reported with a brief review of available literatures to enrich our knowledge of such anomalies in our medical science so that ultimately we can try our best to save the newborn with such anomaly.

In prenatal diagnosis ultrasonography is highly sensitive to detect congenital diaphragmatic hernia. Bowel loops are seen to have peristalsis in the thorax instead of abdomen.[2] In prenatal ultrasonography (USG), detection of polyhydramnios, an absent or intrathoracic stomach bubble; a mediastinal and cardiac shift away from the side of the herniation, and rarely, fetal hydrops might lead to diagnosis of congenital diaphragmatic hernia. In USG, position of the stomach is very important to differentiate CDH from other anomalies like congenital cystic adenomatoid malformation[1] in which large cysts mimicking air-filled intestinal loops are seen in the lungs but here the stomach and the bowel remain normal in position and appearance. Liver herniation might be associated with poorer prognosis detected by radiological investigations in fetal congenital diaphragmatic hernia. Gradation of liver herniation can be used as a marker for the prediction of outcome of fetal congenital diaphragmatic hernia.5 Plain radiography sometimes may be helpful for presumptive diagnosis and contrast CT may help to arrive at a precise diagnosis. As lung tissue perfusion is reduced in CDH so intrapulmonary pulsed Doppler and association with the lung to head ratio might be helpful in prenatal diagnosis of CDH.[6,7]

The baby born with congenital diaphragmatic hernia (CDH) is a surgical emergency situation. If CDH is prenatally diagnosed then to save the life of the baby the mother should be transported to specialized institutes before delivery of CDH newborns. According to Aly H. et al the survival rate in transported newborns was improved with the use of extracorporeal membrane oxygenation (ECMO) and then operated for the repair of CDH. The repair could be done by open or thoracoscopic method. Thoracoscopic repair is more feasible but more study is needed regarding the effects on ventilation and recurrences.[8]

At operation, the posterior rim of the diaphragm is first separated from the overlying peritoneum and then a two-layer closure of the canal is done. Synthetic patch may be needed where the diaphragm is insufficient. To maintain postoperative PaO2 above 100mmHg and a PCO2 less than 40mmHg is very good prognosis. The use of high frequency-ventilation and extracorporeal membrane oxygenation (ECMO) can help in increased survival rate of the severely compromised babies.[9]

CASE STUDY

A new born female baby presented with severe respiratory distress. On clinical examination of the baby scaphoid abdomen and cyanosis was found. Immediate chest X-ray showed an ill-defined non-homogeneous opacity occupying almost whole of the right hemithorax with shift of mediastinum and the heart to the opposite side. The right hemidiaphragmatic silhouette was obscured. CT scan was performed with instillation of contrast through a nasogastric tube.

Scanogram showed contrast filled small intestinal loops in right hemithorax with contralateral shift of the heart and mediastinum. A loop of nasogastric tube was found in stomach under the left hemidiaphragm.Tip of the nasogastric tube entered into the right hemithorax. It was difficult to identify

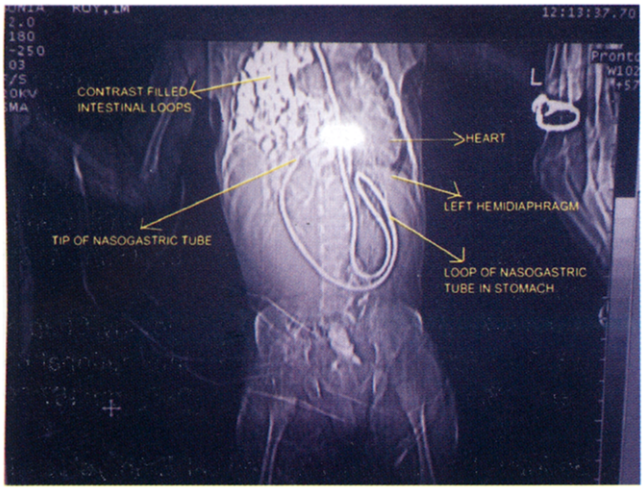

Figure no.1-Scanogram showing contrast filled intestinal loops occupying almost whole of the right hemithorax with contralateral shifting of mediastinum including heart. Nasogastric tube is seen in situ.
Title- Scanogram in supine position-frontal projection.

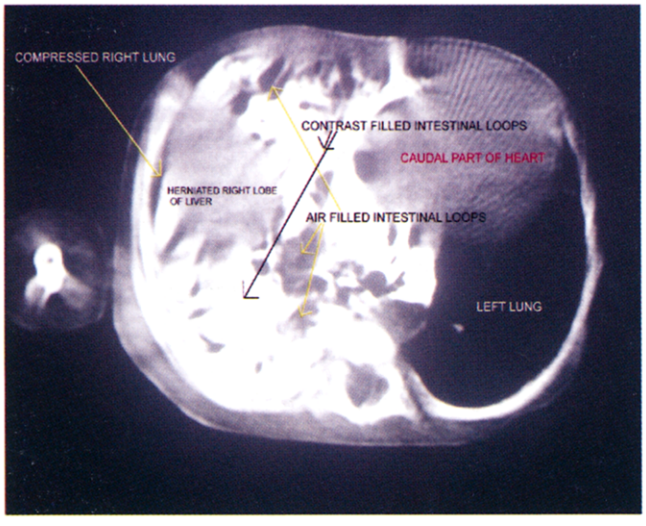

Figure no.2- CT scan of thorax at the level of ventricles showing herniation of contrast filled intestinal loops, part right lobe of liver in the right hemithorax and with peripherally shifted heart in left hemithorax.
Title- CT scan of thorax at the level of ventricles (Mediastinal window setting).

the lung shadow in the right hemithorax in the scanogram, but the left lung was normal (Fig.1). Axial CT scan of thorax at the level of ventricles identified a small and peripherally pushed right lung due to herniation of small intestinal loops and part of right lobe of liver. Contralateral shifting of heart was present (Fig.2).

The baby was delivered by the primae mother (26 year old) with no such obvious abnormalities corroborative with the thorough clinical examination. Karyotype (to detect trisomy, chromosomal deletions etc.) of the baby was normal and family history was negative for congenital diaphragmatic hernia.

EMBRYONIC EXPLANATION

The diaphragm, the mesodermal partition in between thorax and abdomen, is developed from the following sources:

(1)Antero-median part including the central tendon, vena caval and oesophageal openings develop from the septum transversum, which is a sheet of mesoderm lying caudal to the pericardial sac and extends from the ventral body wall to the oesophageal segment of the foregut.

(2)Between the oesophageal and aortic openings, the diaphragm develops from the dorsal mesentery of the oesophagus.

(3)Peripheral part is developed from the shelf-like projection of the body wall.

(4)The pleuro-peritoneal openings are situated dorsal to the septum transversum and on each side of the dorsal mesentery of the oesophagus. Each opening is closed by a pleuro-peritoneal membrane which is dissected off from the body wall by the caudal growth of the lung-bud. The membrane fuses with septum transversum and with the dorsal mesentery of the oesophagus.The right opening closes earlier than the left one[10,11]

Here the defect is in the failure of the right sided pleuro-peritoneal membrane to close the same sided pleuroperitoneal canal. Separation of the pleural and peritoneal cavities is effected by development of the septum transversum. Pleuroperitoneal membranes are dorsolateral to the pleuroperitoneal canals. The pleuroperitoneal canal is closed by the fusion of its edges. Growth of organs surrounding the canal especially suprarenal glands will help to meet the pleuroperitoneal membranes with the pars diaphragmatica of the septum transversum.[11] In the reported case peritoneal and peritoneal cavities are continuous with each other along with posterior body wall as there is a in the development of right sided pleuroperitoneal membrane. The hernia most commonly occurs in the left side as it closes later but here in this reported case the hernia is in the right side which is very uncommon. The diaphragmatic defect allows the abdominal visceras to enter the thoracic cavity. The herniated mass prevents the growth of the right lung causing pulmonary hypoplasia with resultant pulmonary hypertension.[12]

CONCLUSION

Congenital diaphragmatic hernia in newborns is a surgical emergency. In prenatally diagnosed cases of CDH prompt surgical maneuver with other supportive steps can save the lives. The presented case report can enrich our knowledge so that we can save as much as lives possible by taking adequate and prompt necessary steps. Attempt can be taken to treat a prenatally diagnosed case of congenital diaphragmatic hernia by thoracoscopic or fetoscopic endoluminal tracheal occlusion in which there is much scope to do works regarding the ventilation and recurrences.

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