STUDY OF ATYPICAL PRESENTATIONS IN CONGENITAL DIAPHRAGMATIC HERNIAVenkata Ramana Poondla¹, Srilakshmi Kothakoona², K. V. J. Rao³, K. Kameswari⁴**HOW TO CITE THIS ARTICLE:**

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ABSTRACT: AIM: To study the clinical features and outcome of congenital Diaphragmatic Hernias with atypical presentation in Paediatric age group. Children with Eventration of Diaphragm are also included in the present study. **RESULTS:** 20 cases of Diaphragmatic Hernia presented with classical presentation, 4 cases with atypical Presentation and a case with recurrent diaphragmatic hernia. Atypical presentations in congenital Diaphragmatic Hernia are Stomach Volvulus and Malrotation of Midgut with Volvulus Intestine. 3 Cases with atypical presentation succumbed to death. **CONCLUSION:** Congenital Diaphragmatic Hernias are common on left side and carries good prognosis. Cases with atypical clinical presentation have 75% mortality. Right sided Congenital Diaphragmatic Hernias are rare but carries guarded prognosis.

KEYWORDS: Congenital Diaphragmatic Hernia, Diaphragmatic Eventration, Stomach Volvulus, Intestinal Malrotation.

INTRODUCTION: Congenital Diaphragmatic defect embraces congenital diaphragmatic hernia and Diaphragmatic Eventration. It is one of the important causes for Respiratory distress in Newborn and infants, rarely with delayed presentation till childhood. It has an incidence of 1 in 5000 live births.^[1] Advances in Radio Diagnosis facilitated early recognition of the condition in the fetus itself. The earliest English language description of the gross anatomy and Pathophysiology associated with congenital diaphragmatic hernia (CDH) in a newborn was by McCauley, an associate of Hunter, as reported in the Proceedings of the Royal College of Physicians 1754. Until 1980s, the standard of care remained immediate neonatal surgery followed by postoperative resuscitative Care. It is essential to consider that the CDH is a physiologic emergency and not a surgical emergency.

MATERIALS AND METHODS: 25 children of Paediatric age group ranging from 1 day to 5 years with diagnosis of Congenital Diaphragmatic Defects comprising Congenital Diaphragmatic Hernia and Diaphragmatic Eventration are taken up for study. These children were admitted during 2008 to 2014 i.e., over a period of 6 years. Diagnosis was arrived with clinical features, Ultrasonography, Infantogram, Barium Meal Follow through (BMFT) [Figure 1] and CECT with oral contrast are the tools adopted for diagnosis. Repair of diaphragmatic hernia and Plication of Eventration of diaphragm under General Anaesthesia are adopted as treatment modalities. Assisted Ventilation provided for all the cases in the first 72 hours of post-operative period. The laterality of Diaphragmatic Hernia, Eventration of Diaphragm and their age distribution, outcome, complications and mortality are studied. 4 cases with atypical presentation and a case with recurrence of Diaphragmatic Hernia, which was operated outside and referred to the institution, were also included in the present study.

ORIGINAL ARTICLE

RESULTS: Of the 25 cases studied 21 are male children and 4 are female with M: F ratio of 4:1. Majority of the children presented with classical clinical features in the neonatal period (17 cases), 5 cases are infants and 3 cases are of children in the age group of 1-3 year. 4 of the 25 cases presented with atypical presentations like Volvulus of stomach (3 cases) and Malrotation of Gut (1 case). Left Diaphragmatic Hernia [Figure 2] accounts to 16 cases and Diaphragmatic Eventration 9 cases. Left laterality is predominant in both Diaphragmatic Hernia and Eventration of Diaphragm, at a ratio of 21:4. No case of Eventration of Diaphragm on right side was reported. No case of bilateral Congenital Diaphragmatic Hernia was reported in the present study. Of the 6 children succumbed to death 3 are with left diaphragmatic hernia, 2 are with right diaphragmatic hernia and a case with left diaphragmatic Eventration. Overall prognosis is poor for right diaphragmatic hernia. A case of Diaphragmatic Hernia with recurrence was repaired with success. Mortality is 75% in Children with atypical clinical features like Stomach Volvulus and Malrotation of Midgut with Midgut Volvulus survived.

DISCUSSION: The incidence of associated malformations in infants with a CDH ranges from 10% to 50%. Skeletal defects have been noted in as many as 32% of CDH infants and include limb reduction and costovertebral defects. Cardiac anomalies have been found in 24% of infants including rare anomalies like Cantrell's pentology. In one study, 100% of stillborn infants with CDH had associated lethal anomalies. Even in infants who survive to birth but die shortly thereafter, neural tube defects were the most common malformations noted.^[2,3] The diagnosis of a CDH is often made on a prenatal ultrasound (US) examination and is accurate in 40% to 90% of cases. Polyhydramnios has been reported in up to 80% of pregnancies with associated CDH. Three dimensional estimation of the fetal lung volume, calculation of the right lung area to thoracic area ratio, and calculation of the lung to thoracic circumference ratio are three different measurements that may correlate with neonatal outcome, but the lung-to-head ratio has been the most widely used prognostic indicator.^[4,5] The most severely affected infants develop respiratory distress at birth, whereas a majority demonstrates respiratory symptoms within the first 24 hours of life. Most CDH's present in the first 24 hours of life, 10% to 20% of the infants with this defect present later.^[6] The infants presented in later life had recurrent mild respiratory illnesses, chronic pulmonary disease, pneumonia, effusion, empyema, and gastric Volvulus.

The respiratory distress in CDH is due to combination of two factors: uncorrectable pulmonary Hypoplasia and potentially reversible pulmonary hypertension. Atypical presentation is anticipated in CDH when clinical features of Gastric Volvulus.^[7] and Malrotation of Midgut.^[8] are noticed in older children. Recurrent diaphragmatic hernia and small bowel obstruction are the dominant surgical challenges following initial repair. Recurrent hernias may occur in up to 50% of infants undergoing patch repair of the defect and in 10% of primary repairs, and they tend to occur in the first 4 years of life. Operative correction is easily performed through an upper transverse abdominal incision. Laparoscopic and thoracoscopic techniques.^[9] have also been used to repair this defect, but the laparoscopic approach has advantage. Even though Fetal Surgical Correction of CDH is described, the results are not encouraging due to procedure related maternal morbidity and mortality.

CONCLUSION: Majority of cases of diaphragmatic hernia and Eventration of Diaphragm presented with respiratory distress, Mediastinal shift, scaphoid abdomen and bowel sounds in the chest.

ORIGINAL ARTICLE

Atypical presentations in CDH are Malrotation of gut and Volvulus of stomach, which are common in delayed cases of congenital diaphragmatic defects. These atypical cases have high mortality rate.

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<28 days	28 th day to 1 year	1 year
17	5	3

Table 1: Age-wise distribution of cases

Sl. No	Year	Male	Female	Total
1.	2008	2	0	2
2.	2009	3	1	4
3.	2010	4	0	4
4.	2011	5	1	6
5.	2012	3	1	4
6.	2013	2	1	3
7.	2014	2	0	2
	Total	21	4	25

Table 2: Year and Gender wise distribution of Diaphragmatic Hernia cases

	No. of Cases	Good outcome	Mortality
Typical Presentation	21	18	3
Atypical Presentation	4	1	3

Table 3: Outcome of Congenital Diaphragmatic Hernia in Children with Typical and Atypical Presentation

Fig. 1: Barium Meal Follow Through Film of Volvulus Stomach

Fig. 2: Intra-Operative Photo of Left Congenital Diaphragmatic Hernia

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