



CLINICAL PROFILE, MANAGEMENT AND FACTORS ASSOCIATED WITH MORTALITY IN CHILDREN WITH CONGENITAL DIAPHRAGMATIC HERNIA: A RETROSPECTIVE STUDY

Paediatrics

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ABSTRACT

Background: Congenital diaphragmatic hernia (CDH) is a major congenital anomaly characterized by a defect in the diaphragm with herniation of abdominal viscera into the thoracic cavity. Pulmonary hypoplasia, pulmonary hypertension, associated congenital anomalies and the severity of cardiopulmonary compromise contribute substantially to morbidity and mortality. Advances in antenatal diagnosis, neonatal stabilization, pulmonary hypertension management and surgical care have improved outcomes; however, severe disease continues to be associated with significant mortality. **Aim:** To describe the clinical profile, management and outcomes of children with congenital diaphragmatic hernia and to identify clinical factors associated with mortality. **Methods:** A retrospective observational study was conducted from period of July 2021 to July 2026 using available clinical data of patients diagnosed with congenital diaphragmatic hernia. Children under the age of 16 who were radiologically diagnosed with congenital diaphragmatic hernia and underwent management in our hospital were included in the study. Participants who underwent surgical intervention in an outside hospital, and individuals with incomplete or missing clinical data were excluded from the study. Demographic, antenatal and postnatal diagnostic characteristics, side of the defect, pulmonary hypertension, associated congenital anomalies, preoperative intubation, surgical intervention, ventilatory support, NICU stay and postoperative complications were analyzed. Continuous variables were summarized using mean $\pm$ standard deviation and median with interquartile range. Associations between categorical variables and mortality were assessed using Fisher's exact test. Kaplan–Meier survival analysis was performed for patients with available follow-up data. A two-sided p-value <0.05 was considered statistically significant. **Results:** Twenty-five patients were included in the available clinical dataset. The majority were male (68.0%). Antenatal diagnosis was documented in 60.0% of patients, while 32.0% were diagnosed during infancy and 8.0% at 2–3 years of age. Left-sided CDH predominated (95.7%). Pulmonary hypertension was documented in 80.0% of patients, and patent ductus arteriosus in 68.0%. Preoperative intubation was required in 86.4% of patients for whom this information was available. Surgery was documented in 76.0% of the cohort. Sixteen patients required ventilatory support and 14 had a recorded NICU stay. Among patients with available mortality data (n=23), six patients (26.1%) died, and 17 (73.9%) survived. Significant associations with mortality were observed for the recorded surgery variable (p=0.002), ventilator requirement (p<0.001), NICU stay (p<0.001), post-operative complication (p=0.001) and lung-related complications (p=0.037). Gender, timing of diagnosis, gestational age, side of defect, pulmonary hypertension, ASD, PDA, pulmonary hypertension medication, herniated contents, preoperative intubation, sepsis, AKI, neurological complications, and other complications like anemia, GI complications were not statistically significantly associated with mortality. Kaplan–Meier analysis included 23 patients, with six deaths. The estimated mean survival time was 38.4 days (SE 4.41 days); the median survival time was not reached during the observed follow-up period. **Conclusion:** This cohort demonstrated a substantial burden of cardiopulmonary morbidity and a mortality rate of 26.1% among patients with available mortality data. Mortality was significantly associated with several recorded clinical variables. The findings emphasize the importance of early diagnosis, optimized neonatal stabilization, pulmonary hypertension management and multidisciplinary perioperative care. Larger prospective studies are required to identify independent predictors of mortality.

KEYWORDS

Congenital Diaphragmatic Hernia; CDH; Pulmonary Hypertension; Neonatal Surgery; Ventilation; Mortality; NICU; Congenital Anomaly

INTRODUCTION

Congenital diaphragmatic hernia is a potentially life-threatening congenital anomaly resulting from defective development of the diaphragm and abnormal communication between the thoracic and abdominal compartments. The clinical severity of CDH is determined not only by the size and anatomical characteristics of the diaphragmatic defect but also by the degree of pulmonary hypoplasia, pulmonary hypertension, cardiac function and associated congenital anomalies.

Contemporary management of CDH has evolved substantially from immediate operative repair after birth toward a strategy emphasizing physiological stabilization before surgical intervention. Current approaches include gentle ventilation, avoidance of excessive airway pressures, treatment of pulmonary hypertension, cardiovascular support and individualized timing of repair. Recent consensus recommendations have further emphasized standardized prenatal assessment, risk stratification and multidisciplinary management. [2,8,11,19,20]

Prenatal diagnosis provides an opportunity for risk assessment and counselling. Several antenatal parameters have been evaluated to estimate disease severity, including defect size, lung size and liver position. More recently, MRI-derived measurements and other quantitative parameters have been investigated to improve prediction of outcome. [5,12]

Pulmonary hypertension remains one of the most important determinants of morbidity in CDH. Persistent pulmonary vascular abnormalities may result in right ventricular dysfunction, impaired oxygenation, and prolonged ventilator dependence. Cardiac dysfunction may also contribute to poor outcomes. [11,13,14]

The anatomical management of the diaphragmatic defect has also evolved. Open repair remains widely used, while thoracoscopic repair has increasingly been adopted in selected neonates. However, minimally invasive repair must be considered in the context of physiological stability, and patient selection remains important. [3,7]

For severe fetal CDH, fetal endoscopic tracheal occlusion has emerged as an antenatal intervention intended to promote lung growth. Randomized trials have demonstrated benefit in carefully selected severe left-sided CDH, although fetal intervention requires specialized expertise and careful multidisciplinary selection. [15-17]

Long-term follow-up is increasingly recognized as an important component of CDH care because survivors may develop recurrent hernia, respiratory morbidity, gastrointestinal problems, neurodevelopmental issues and other long-term complications. [4]

The present study was undertaken to describe the clinical profile, management and outcomes of patients with CDH treated at the study center and to evaluate clinical factors associated with mortality.

MATERIALS AND METHODS

Study Design and Setting

This was a retrospective observational study based on clinical data of patients diagnosed with congenital diaphragmatic hernia and managed at the study center.

Study Population

All patients diagnosed with congenital diaphragmatic hernia and managed at the study center during the study period January 2021 to July 2026 were considered for inclusion. Patients were included when clinical, operative, and outcome information was available in the study records. The available dataset contained 25 patients for most baseline variables, while mortality and survival information was available for 23 patients.

Variables Studied

The study proforma included demographic characteristics, timing of diagnosis, radiological findings, associated congenital abnormalities and perioperative variables.

- Age
- Gender
- Timing of diagnosis
- Gestational age
- Side of diaphragmatic defect
- Pulmonary hypertension
- Associated congenital defects, including ASD
- Patent ductus arteriosus
- Use of medication for pulmonary hypertension
- Herniated abdominal contents
- Preoperative intubation
- Surgical intervention
- Requirement for ventilatory support
- NICU stay
- Postoperative complications (All complications included)
- Sepsis
- Lung-related complications
- Acute kidney injury
- Neurological complications
- Other complications – hematological, gastrointestinal
- Duration of hospitalization
- Birth weight
- Day of surgery
- Duration of ventilator support
- Time to initiation of oral feeds
- Duration of NICU stay
- Mortality

Statistical Analysis

Data were analyzed using IBM SPSS Statistics version 23.0. Categorical variables were expressed as frequencies and percentages. Continuous variables were summarized using mean ± standard deviation and median with interquartile range.

Because of the relatively small sample size and low expected cell frequencies, Fisher's exact test was used to assess the association between categorical variables and mortality. A two-sided p-value < 0.05 was considered statistically significant.

Kaplan–Meier survival analysis was performed among patients with available survival data. Survival time was calculated from the date of birth to the date of death or the last documented follow-up, as applicable. Patients alive at the end of follow-up were treated as censored observations. Mean survival time was reported with its standard error. The median survival time was considered not estimable when fewer than 50% of patients had experienced the event during the available follow-up period.

RESULTS

Demographic and Clinical Characteristics

A total of 25 patients were represented in the baseline dataset. There was a male predominance, with 17 males (68.0%) and 8 females (32.0%). Referred out patients: 2/25 (8.0%).

Antenatal diagnosis was documented in 15 patients (60.0%). Eight patients (32.0%) were diagnosed during infancy, while two patients (8.0%) were diagnosed at 2–3 years of age.

Among patients with available gestational-age information, 20 (87.0%) were term and three (13.0%) were preterm. The defect was

predominantly left-sided, occurring in 22 patients (95.7%), while one patient (4.3%) had a right-sided defect.

Pulmonary hypertension was present in 20 patients (80.0%). Associated ASD was recorded in 15 patients (65.2%) in the available dataset, while PDA was documented in 17 patients (68.0%). Medication for pulmonary hypertension was used in 17 patients (68.0%). Herniated abdominal contents were recorded in 11 patients (44.0%) (Table 1).

Table 1. Baseline Clinical Characteristics. Denominators Vary Because of Missing Observations in the Source Dataset.

Variable	n	%
Gender – Male	17	68.0
Gender – Female	8	32.0
Timing – Antenatal	15	60.0
Timing – Infancy	8	32.0
Timing – 2–3 years	2	8.0
Gestational age – Term	20	87.0
Gestational age – Preterm	3	13.0
Side – Left	22	95.7
Side – Right	1	4.3
Pulmonary hypertension – Present	20	80.0
Pulmonary hypertension – Absent	5	20.0
PDA – Present	17	68.0
PDA – Absent	8	32.0
PAH medication – Used	17	68.0
PAH medication – Not used	8	32.0
Herniated contents – Present	11	44.0
Herniated contents – Absent	14	56.0

Perioperative Characteristics and Complications

Preoperative intubation was documented in 19 of 23 patients (82.6%) for whom this variable was available. Surgical intervention was recorded in 19 patients (76.0%). Sixteen patients (64.0%) required ventilatory support, and 14 (56.0%) had a recorded NICU stay.

The dataset recorded 14 patients (56.0%) with postoperative complications, which include 7 patients (28.0%) with sepsis, lung-related complications in nine patients (36.0%), and AKI and neurological complications were each recorded in four patients (16.0%). Other complications, such as hematological and gastrointestinal complications, were present in five patients (20.0%).

Table 2. Perioperative Characteristics and Complications. *Denominator for Preoperative Intubation = 22.

Variable	n	%
Preoperative intubation*	19	82.6
Surgical intervention	19	76.0
Ventilator support	16	64.0
NICU stay	14	56.0
Post operative complications	14	56.0
Sepsis	7	28.0
Lung-related complications	9	36.0
Acute Kidney Injury	4	16.0
Neurological complications	4	16.0
Other complications	5	20.0

Continuous Variables

The mean duration of hospitalization was 18.35 ± 13.87 days, with a median of 13 days (IQR 8–30). Mean birth weight was 2.61 ± 0.60 kg, with a median of 2.60 kg (IQR 2.24–3.00).

The mean day of surgery was 3.21 ± 1.65 days, with a median of 3 days (IQR 2–4). Among patients with available data, mean duration of ventilator support was 8.19 ± 5.48 days, and the mean time to oral feeding was 6.69 ± 4.98 days. Mean NICU stay was 20.43 ± 14.96 days.

Table 3. Continuous Clinical and Outcome Variables.

Variable	N	Mean ± SD	Median (IQR)
Duration of hospitalization (days)	17	18.35 ± 13.87	13 (8–30)
Birth weight (kg)	23	2.61 ± 0.60	2.60 (2.24–3.00)
Day of surgery	19	3.21 ± 1.65	3 (2–4)
Days on ventilator	16	8.19 ± 5.48	7 (3–11.75)

Days till oral feed	16	6.69 ± 4.98	5.5 (4–8)
NICU stay (days)	14	20.43 ± 14.96	15.5 (12–28)

Mortality

Mortality data were available for 23 patients as two patients went left to other centers for further management due to their personal convenience. Six patients (26.1%) died, whereas 17 (73.9%) survived during the available observation period.

Table 4. Mortality in the Study Cohort.

Outcome	n	%
Survived	17	73.9
Died	6	26.1
Total (Excluding referred patients)	23	100

Table. Summary of Study Population and Outcomes

Parameter	Number of Patients	Percentage (%)
Study population	25	100.0
Referred patients	2/25	8.0
Overall mortality	6/23	26.1
Survivors	17/22	73.9

Note: The denominators for mortality (23) and survival (22) differ. Also, 17/22 equals 77.3%, whereas 17/23 equals 73.9%. These values should be verified before finalizing the table.

The following is a summary based on the cause-of-death notes provided. The categories overlap because individual patients may have had multiple complications. They should not be interpreted as mutually exclusive causes of death until verified against each patient's records.

Reported cause or complication	Number Identified in Notes	Percentage of Total Patients (N=25)
Deaths from all causes	6	24%
Hemorrhage with hypovolemic shock / cardiac arrest	2	8%
Sepsis	2	8%
Renal dysfunction / acute kidney injury (AKI)	3	12%
Recurrent bradycardia	1	4%
Respiratory acidosis	1	4%

DISCUSSION

Congenital diaphragmatic hernia (CDH) remains a complex neonatal condition whose outcome depends on the interaction between pulmonary hypoplasia, pulmonary vascular disease, cardiac function, associated anomalies, and postnatal physiological responses. In this cohort, left-sided defects predominated (95.7%), consistent with established clinical series. Antenatal diagnosis was achieved in 60% of patients, offering critical opportunities for foetal risk stratification, counselling, and delivery planning. The high prevalence of pulmonary hypertension (80%), preoperative intubation (86.4%), and mechanical ventilation (64%) underscores the heavy cardiopulmonary burden and persistent respiratory compromise inherent to CDH.

Overall mortality in the cohort was 26.1%, with an estimated mean survival time of 38.4 days. Lung-related complications were significantly associated with mortality (p=0.037), highlighting the central role of underlying airway and vascular abnormalities in patient outcomes. Conversely, while pulmonary hypertension, ASD, PDA, and specific medications were not statistically linked to mortality, these non-significant p-values must be interpreted cautiously due to low statistical power in a small sample with only six deaths.

Modern CDH management continues to emphasize physiological stabilization before delayed surgical repair, reserving thoracoscopic approaches for carefully selected stable neonates. Antenatal therapies such as foetal endoscopic tracheal occlusion (FETO)—supported by the TOTAL trial—provide specialized options for severe left-sided cases. Finally, given the risks of defect recurrence and long-term sequelae, structured, multidisciplinary follow-up remains essential throughout childhood.

Limitations

This study has several limitations.

1. The sample size was small, with 25 patients represented in the baseline dataset and mortality data available for only 23 patients. The small number of deaths substantially limits statistical power.
2. The retrospective nature of the study introduces the possibility of

- missing data and information bias. This is reflected by variable denominators across the analysis.
3. Only univariate categorical associations using Fisher's exact test were performed. Multivariable regression analysis was not undertaken. Therefore, statistically significant variables cannot be considered independent predictors of mortality.
4. The available Kaplan–Meier analysis provides an estimate of survival during the observed period but does not establish long-term survival or long-term morbidity.

CONCLUSION

This retrospective cohort demonstrates the substantial clinical burden associated with congenital diaphragmatic hernia, with a high frequency of pulmonary hypertension, respiratory support requirements and associated complications. Mortality was 26.1% among the 23 patients with available mortality data.

Several variables, including surgery, ventilator requirement, NICU stay, and lung-related complications, were statistically associated with mortality. However, these findings should be interpreted cautiously because of the small sample size and potential coding inconsistencies.

The findings reinforce the importance of early diagnosis, careful antenatal risk assessment, optimized neonatal stabilization, management of pulmonary hypertension, multidisciplinary intensive care and appropriately timed surgical repair. Larger prospective studies with standardized definitions and complete longitudinal follow-up are required to establish independent predictors of mortality and long-term outcomes in children with CDH.

Declarations

Ethics Approval

Ethics committee approval has been obtained according to the institutional protocol.

Consent

Consent was waived off by the institutional ethical committee keeping in view of retrospective observational study.

Conflict of Interest

No conflict of interest by any author.

Funding

No funding was obtained.

Data Availability

The data supporting the findings of this study was available from the corresponding author subject to institutional policies and ethical restrictions.

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