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Congenital diaphragmatic hernia in neonate: A retrospective study about 28 observations

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ABSTRACT

Objective: Our purpose was to review our experience with congenital diaphragmatic hernia emphasizing diagnosis, management, and outcome. **Study Design:** We conducted a retrospective review of all cases of babies with congenital diaphragmatic hernia diagnosed and treated in our centre from 1998 to 2010. **Results:** There were 28 congenital diaphragmatic hernia cases, 13 girls and 15 boys with a mean weight birth of 3 kg. Three patients (10, 6% of cases) died within a few hours after admission. In the remaining cases, surgery was performed after a stabilization period of 2 days. The diaphragmatic defect was sitting in the posterolateral left in 23 cases and right in 2 cases. Its dimensions were on average 4,5 cm, tow cases of agenesis of the cupola were seen and required the placement of gortex prosthesis. The remaining cases are treated by direct closure of defect. Postoperative course was marked by an early death in context of respiratory distress in six cases and later with sepsis in tow cases. The outcome was favourable in 17 cases (60, 7%), despite the occurrence of sepsis in four cases and evisceration in two cases. **Conclusions:** Congenital diaphragmatic hernia remains a serious disease with high mortality and morbidity despite advances in prenatal diagnosis and neonatal resuscitation.

Key words: Congenital diaphragmatic hernia, pulmonary hypoplasia, tracheal occlusion, paediatric surgery

Congenital diaphragmatic hernia (CDH) is a frequent malformation characterized by the existence of a diaphragmatic defect located mostly in posterolateral side through which abdominal viscera will migrate into the chest.

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Apart from the late-onset forms in which the prognosis is generally good, the congenital diaphragmatic hernia is a very serious malformation.

In recent years, the management of CDH has evolved through prenatal diagnosis and especially the new techniques of neonatal resuscitation. Despite these developments, prognosis remains charged with a heavy short-term mortality and high morbidity in medium and long term.[1]

The aim of our study is to report the experience of paediatric surgery department of the Children's Hospital of Tunis in the management of this malformation in a 13-year period and to compare these results with those of the literature.

MATERIALS AND METHODS

Over a period of 13 years (1998–2010), 28 newborn babies with a congenital diaphragmatic hernia have been managed in the Department of Pediatric Surgery "B" at the Children's Hospital of Tunis. There are 13 girls and 15 boys with a sex ratio of 1:15.

Twenty-four of our patients were born at term, while the remaining four patients were premature with an average term of 33 weeks. The mean birth weight was 3 kg and ranged from 1100 to 4100 grams.

In five cases, the diagnosis was established on prenatal obstetrical ultrasound performed in the third trimester, while in the remaining cases the diagnosis was made at birth during a respiratory distress in the first day in 18 cases, or arising in the first week of life in five cases [Table 1].

The diagnosis was made on the chest radiograph which showed digestive lights in the left hemithorax in 24 cases and in the right hemithorax in three cases. In one case, a clouding of the left pulmonary base made the diagnosis suspected [Figures 1, 2]. In three observations,

an opacification of the upper gastrointestinal tract through a gastric tube was performed to confirm diagnosis [Figure 3].

In 20 cases, the respiratory conditions were so severe that it required intubation with mechanical ventilation. In the eight remaining cases, homeostasis was provided by an oxygenation to hood.

Table 1: Clinical aspect of population

Diagnosis	
Antenatal	5
First day respiratory distress	18
First week respiratory distress	5
Sex	
Boy	15
Girl	13
Term	
Full term	24
Preterm	4

Three patients (10,6% of the series) died within a few hours after admission despite resuscitation. In the other cases, surgery could be performed after a stabilization period of 2 days on average.

The intervention was conducted by a left subcostal incision in 17 cases, right transverse incision in 2 cases, and median laparotomy in the 6 remaining cases.

The diaphragmatic defect was sitting in the posterolateral left in 23 cases and right in 2 cases [Figure 4]. Its dimensions were on average 4, 5 cm, tow cases of agenesis of the cupola were seen and required the placement of gortex prosthesis. In 4 cases (22.2% of cases), there was a hernia sac and in 16 cases a poorly mesenteric rotation.

It made a reduction of herniated organs, excision of the hernia sac, and a freshening of the banks with direct closure of diaphragmatic defect with no absorbable suture without drainage. The associated malrotation

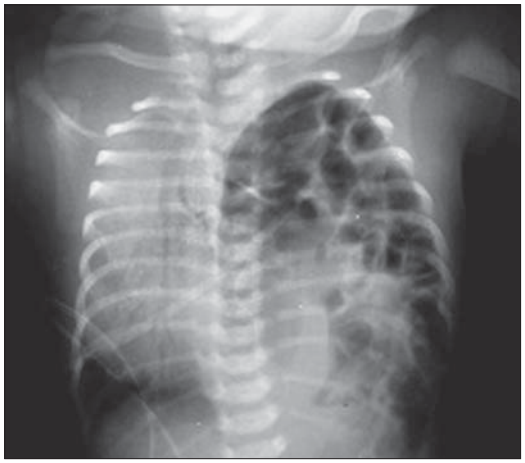


Figure 1: Chest radiography showing digestive lights in the left hemithorax



Figure 2: Chest radiography profile showing digestive lights in the left hemithorax

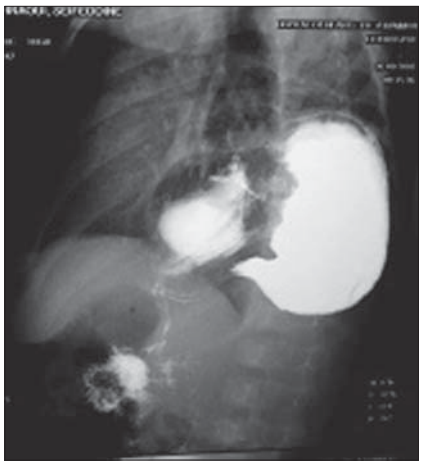


Figure 3: Gastro intestinal opacification showed intrathoracic stomach

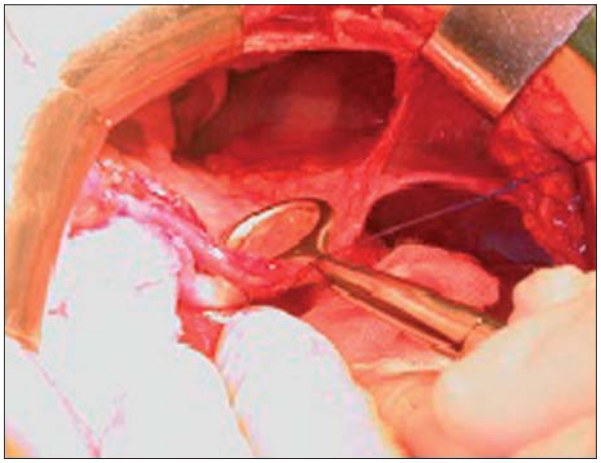


Figure 4: Peroperative view: large diaphragmatic defect with pulmonary hypoplasia

was treated by setting bowel loops in common complete mesentery with appendectomy [Table 2].

All patients were kept intubated and ventilated postoperatively for an average of 6 days and 6 hours with a range from 1 to 32 days. The evolution was marked by an early death despite the use of nitric oxide and high-frequency ventilation in six cases (first week) and late (D30PO, D15PO) in a context of septicemia in tow patients.

The outcome was favourable in 17 cases (60,7%), despite the occurrence of sepsis that requires prolonged antibiotic therapy in 4 cases and evisceration that required further surgery in 2 cases.

From our four premature patients, two died in preoperative period and two in postoperative period. And one of the babies with right-sided defect is dead in postoperative days [Table 3].

The poor prognostic factors identified were low birth weight, the large size of defect, the location of right diaphragmatic defect, and associated cardiac anomalies.

DISCUSSION

The CDH occurs in 1/2000 pregnancies and 1/3000 live births, reaching especially the girls with a sex ratio of 3 girls 2 boys (in our series the sex ratio is 1:15).[2]

Familiar CDH has been reported, and it has been estimated that these comprise about 2% of all cases.

Table 2: Operative findings

Surgical approach	
Left subcostal	17
Median	6
Right transverse	2
Diaphragmatic defect site	
Left posterolateral	23
Right posterolateral	2
Surgical procedure	
Direct closure	23
Prosthesis	2

Table 3: Evolution of patients

Evolution	Preoperative	Postoperative		Total
	Death	Favourable	Death	
Premature	2	0	2	4
Right-side defect	1	1	1	3
Others	0	16	5	21
Total	3	17	8	28

Most of the reported cases have involved posterolateral defects. The mode of inheritance of familial CDH is speculative. Early reports suggested an autosomal recessive pattern, but other authors do not agree with this.[3] CDH seems to be mostly sporadic. No familial form has been encountered in our series.

Despite advances in diagnosis and technical care, the neonatal revelation of CDH raises serious problems of management and presents a heavy mortality and morbidity. The condition may be life-threatening and mortality rates in live-born patients range from 10% to 35%. The true mortality may even be higher when taking into account antenatal death or termination of pregnancy. Key determinants of mortality are the severity of pulmonary hypoplasia and the presence of pulmonary hypertension.[1,4,5]

The most common HCD is posterolateral hernia of Bochdalek, typically 2–3 cm diameter, on the left in 75 to 85% of cases. Sometimes the fault is wider, bordering on diaphragmatic agenesis more or less complete.[6] The bilateral form is rare and usually associated with other birth defects. In 15–20% of cases, hernia sac is present and a partition already made between the chest cavity and abdomen is witnessed.

The diaphragmatic hernia accompanied by associated malformations, which have an impact on the pejorative prognosis; they are very common in stillbirths (85%) and would range from 20% to 50% among children born alive but much of which cannot until surgery, thus explaining their relative scarcity in surgical series.[1]

They affect different devices: Central nervous system (30%), with a particular incidence of anencephaly, genitourinary and cardiovascular system. The frequency of cardiovascular malformations (CVMs) associated with isolated CDH was 11–15%. A careful analysis of them indicates that atrial and ventricular septal defects, conotruncal defects, and left ventricular outflow tract obstructive defects were the most common type of CVMs. The combination of CVMs and CDH results in a poorer prognosis than would be expected with either malformation alone.[7]

Chromosomal abnormalities are not uncommon (3% among live births) and justify amniocentesis during antenatal diagnosis. Aneuploidies, genetic syndromes, and structural abnormalities of chromosomes, such as deletions, duplications, inversions, and translocations, may indicate the involvement of genetic factors. The most reported aneuploidies in CDH patients are trisomy

13, 18, 21, and 45X, and the most reported genetic syndrome is Fryns syndrome.[5] Other anomalies have been reported such as pulmonary sequestration (0.7% of cases), esophageal atresia.

The colonization of a hemithorax by abdominal viscera causes compression of the lung parenchyma and may result in more serious cases to severe pulmonary hypoplasia which is constant in the very early-onset forms and then dominate the prognosis. This concerns both the ipsilateral and the controlateral lung.

The diagnosis of diaphragmatic hernia may be suspected prenatally or done in postnatal period in front of acute respiratory distress

Prenatal diagnosis is based on obstetrical ultrasound; it is more difficult on the right side. According to the different teams and/or extent of the hernia, gestational age at diagnosis ranges from the 15th to the 41st week of gestation.

More detailed ultrasound techniques allow for prenatal detection of CDH in almost 60% of the cases. Several prenatal predictors have been proposed to determine postnatal chances of survival. These predictors may also be used to determine the need for fetal surgery.[5]

Adzick and Coll consider a prenatal diagnosis before 25 weeks is a poor prognosis. This traditional concept tends to disappear from the recent literature. Indeed, the authors concur that the profitability of the ultrasound is improving prenatal diagnosis allowing more early and then a fetus, some of which are carriers of lethal malformations.[8]

The diagnosis of HCD justifies the obtaining of a fetal caryotype, because as we have already seen it is accompanied by genetic abnormalities that may influence the prognosis for survival and support postnatal. It will be established by examining the fibroblasts in order not to miss an anomaly in mosaic, such as trisomy 12p, or Pallister Killian syndrome.

In the absence of prenatal diagnosis, the HCD is manifested in the first hours of life by acute respiratory distress, which may be immediate or gradual.

Radiological examination is an emergency, a thoracoabdominal radiogram face often enough to make the diagnosis by showing intrathoracic multiple gas clearness associated with deviation of the heart and mediastinum to the right side and declining levels of digestive pneumatization in the abdomen.

The radiological diagnosis is sometimes difficult and was the case of right diaphragmatic hernia where the defect is filled by the liver, and if the radiograph is made early before the intestinal pneumatization shows an opaque base of the thorax posing the problem of differential diagnosis.

Predicting survival, either in affected fetuses detected on antenatal ultrasound or live-born infants, is challenging; numerous studies have been attempted. However, no predictor appears to have been generally accepted into clinical practice. Recently, direct assessment of the fetal lungs has appealed as a marker for lung hypoplasia, the most significant determinant of survival. The most standardized measurement for lung hypoplasia is the lung-to-head ratio (LHR), which has been accepted by most groups. It is obtained by dividing the contralateral lung area to the fetal head circumference.[4,5,9] This ratio was excluded from our survival analysis because the rate of outborn delivery without prenatal diagnosis was very high at our institution.

Recently, the fetal lung volume has been proposed as a prenatal predictor of survival in fetuses with congenital diaphragmatic hernia.[5] Among the clinical characteristics, birth weight and Apgar score at 5 min were found to be the most important predictors to estimate the severity of CDH in the first 5 min of life by the Congenital Diaphragmatic Hernia Study Group.[2] Moreover, the outcomes for preterm infants are clearly worse than in the term infant, and preterm infants with CDH remain a high-risk group as well as the right side of the defect.[10] This was confirmed in our series.

For over two decades, efforts have been deployed to rescue lung growth in the severely affected fetus with CDH. The work of Harrison's group in San Francisco initially focused on open antenatal repair of the diaphragmatic defect. But outcomes were marked by problems maintaining tocolysis and a high rate of preterm delivery.

Further advances resulted from the key observation that babies born with congenital laryngeal atresia develop tremendous pulmonary growth. This finding prompted the concept that occluding the fetal trachea in pulmonary hypoplasia associated with CDH could increase lung growth and improve outcomes. Initial attempts at open tracheal occlusion were succeeded by a modified endoscopic technique. However, significant variability in lung growth has been observed in early clinical trials of tracheal occlusion.[11,12] This may be caused by the fact that lung hypoplasia created at relatively late stages of lung development may not be

equivalent to human CDH-induced lung hypoplasia, which begins early in gestation.[11]

A randomized trial led by the San Francisco group disappointingly showed no survival benefits compared with elective delivery at specialist centres with optimal postnatal CDH care.[13] In Europe, fetal tracheal occlusion (FETO) continues to be explored, and further technical refinement may uncover survival benefits in high-risk patients.[4]

In case of prenatal diagnosis, birth is required in a reference centre Level III, where a team of paediatric surgeons and neonatal resuscitation can provide a rapid response of the newborn. There is no preference for a particular mode of delivery; it should be as close to term. In the case of an unexpected birth, transportation must be provided by a team of paediatric Department Mobile of Emergency and Reanimation.[1]

For decades, the CDH has been supported as a major neonatal surgical emergency. The child quickly conditioned and operated within hours of birth. In the mid-80s, the concept of preoperative stabilizing has gradually been adopted.[1,4,5,14] This approach permits full assessment and stabilisation of labile physiology. Increasingly, cardiologists can play a pivotal role in serial echocardiographic assessment to monitor ductal shunting and pulmonary hypertension. A team approach is crucial to guide the optimum timing of surgery.[4]

The means of preoperative stabilization are as follows:

- Conventional ventilation using fast frequency between 80 and 150/min.
- High-frequency oscillation with a recognized quality to minimize barotrauma by avoiding the plateau pressure source of alveolar rupture and pneumothorax.
- Extracorporeal membrane oxygenation that provides a blood oxygenation and decarboxylation by a continuous exchange of oxygen and CO_2 between an extracorporeal blood circuit and a circuit gas separated by a membrane permeable only to gas. Two modes are used: the venoarterial and venovenous techniques.[1,4] This technique is reserved for specialized centers. In our country we do not have this method of management.
- Inhaled nitric oxide which causes selective vasodilation of the pulmonary vessels, reverses the right-left shunt and correct the hypoxemia. Unfortunately, the neonates with CDH respond inconstantly to this treatment, and pulmonary vascular resistance is fixed because of anatomical reduction of the vascular lumen.[2,4,5]
- Exogenous surfactant: Wilcox *et al.* experimented instillation of surfactant into the trachea of fetal lambs carrying a CDH and compared their ventilatory parameters with control cases. Lung volume, Functional residual capacity and compliance were all significantly improved.[15] These results were not confirmed by a study performed in the fetus to know whether there is a deficit in surfactant or not. These results indicate that CDH does not impair surfactant storage in human fetuses and lungs exhibited no trend towards a decrease in contents, or a delay in developmental changes for any of the studied surfactant components and surfactant maturation factors. Surfactant amounts are likely to be appropriate to lung size.[16] These findings therefore do not support the use of surfactant therapy for infants with CDH. Moreover, they raise the question of the relevance of CDH animal models to explore lung biochemical maturity.[16]

Surgical management is often performed by laparotomy via a subcostal incision ipsilateral to the hernia. Herniated viscera are reduced, and the diaphragmatic gap is visualized and lung volume assessed. The closure of the diaphragmatic defect is performed either by suture or by using a plastic surgery[4] depending on the size of the orifice and the existence of a posterior diaphragmatic rim, which is the pledge of solidity. This surgery can be made by laparoscopic approach especially in older children with a delayed presentation.[17-19] The question that remains discussed is the establishment of a prosthetic diaphragmatic, and some authors believe that it improves lung compliance in case of large defect and others consider that there is a risk of recurrence of the hernia.

Comparisons of survival rates of CDH patients between institutions or periods are difficult and controversial because the spectrum of disease severity is very wide. In recent retrospective reviews of CDH cases, overall survival rates ranging between 50% and 56% were observed. This rate seems higher in North American institutions.[2]

Close attention to CDH survivors reveals a high incidence of long-term morbidity, particularly focused on the respiratory and gastrointestinal systems.[1,4]

CONCLUSION

Despite the considerable work that this malformation has raised, the mortality rate of congenital diaphragmatic hernia remains very high. It is necessary to use in a rational way the therapeutic resources available to end to improve both survival and reduce long-term sequelae.

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