CONGENITAL DIAPHRAGMATIC HERNIA: A RETROSPECTIVE AUTOPSY STUDY

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ABSTRACT

Congenital diaphragmatic hernia is one of the acute pediatric surgical emergencies and an important cause of perinatal mortality. Its incidence varies from 1 in 2000 to 1 in 5000 live-births, with a greater incidence in stillbirths and abortions. Non-operative mortality is generally reported as 100% and early diagnosis is imperative. A retrospective study of all infant and neonatal autopsies done during last 30 years (1960 to 1989) was conducted. Ten cases of congenital diaphragmatic hernia were encountered among 588 autopsies and its incidence was 1.7% of all infant and neonatal autopsies, dying due to varying causes. The male:female ratio was 7:3, while maternal factors showed no consistent relationship. All cases had left-sided posterolateral diaphragmatic defects. Dyspnea, cyanosis and dextrocardia was the classical triad present in more than 50% cases. The commonest herniated contents were the small intestinal loops and the left lobe of liver. Associated congenital multiple anomalies related to neural axis, skull, vertebral column and cardiovascular system were noted in 30% cases and were major contributory factors for perinatal mortality.

Key words: *Congenital diaphragmatic hernia, Autopsy.*

Congenital diaphragmatic hernia, till today continues to be a critical problem for neonatal survival. Despite the apparent simplicity of the anatomic defect, the physiology is complex, and survival remains uncertain. New and apparently effective combined investigative and surgical regimens have, therefore, recently become available for patients with this defect. Hence, it becomes increasingly important that appropriate parameters be established which are able to predict with accuracy which patients are likely to benefit from antenatal diagnosis, to determine whether they might be reversible, as well as those for whom surgical intervention is imperative. To that end, we have evaluated our experience with 10 cases of congenital diaphragmatic hernia, accumulated retrospectively over a 30-year period in the Department of Pathology, Lady Hardinge Medical College. Some potentially useful clinicopathologic and autopsy findings are presented.

Material and Methods

The files of the Pathological Autopsy Division of the Department of Pathology were reviewed over a 30-year period (1960-1989). Of a total of 588 autopsies conducted, on infants and neonates dying due to varying causes, 10 cases of congenital diaphragmatic hernia were encountered. The patients' records were evaluated in each case for epidemiological aspects,

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maternal and fetal clinical details, and gross and microscopic pathology.

Results

Of the ten neonatal autopsies studied, 8 were livebirths and 2 stillbirths. Of 8 livebirths, 7 (87.5%) neonates died within the first 8 hours of birth, while 1 (12.5%) died 37 hours after birth. The male:female ratio was 7:3. There were 7 fullterm and 3 preterm infants. Of the preterms, 2 neonates had birth weights of 2.0 kg, while 1 neonate weighed 2.2 kg (*Table I*).

Of the 10 infants, 7 had normal vaginal delivery, 2 were delivered by forceps and 1 by cesarean section. There were 4 primigravida and 6 multigravida mothers; 1 had eclampsia of pregnancy while 9 mothers had uncomplicated pregnancy (*Table I*).

All the 8 livebirths showed low and deteriorating Apgar scores at birth. Dyspnea and cyanosis developed within 2-6 minutes of birth in 5 cases. Dextrocardia with displaced apex beat was seen in all 8 livebirths. Other accompanying clinical features were; scaphoid abdomen (37.5% livebirths), diminished breath sounds (62.5% livebirths), increased percussion dullness on affected side (37.5% livebirths) and bowel sounds audible in the chest (37.5% livebirths) (*Table I*). Chest roentgenogram, available in 1 case showed presence of bowel loops within the thoracic cavity.

An accurate clinical diagnosis of congenital diaphragmatic hernia at birth was possible in only 6 (60%) of the total 10 cases. The spectrum of clinical diagnoses in the rest 4 cases was: prematurity; severe birth asphyxia; birth asphyxia with dextrocardia; and preterm with ruptured meningomyelocele.

All the 10 neonatal autopsies studied, showed left-sided posterolateral defects (*Fig. 1*). There was no case of bilateral hernia. The commonest herniated contents were: intestinal loops (80%); left lobe of liver (50%); stomach (40%) and spleen (30%) (*Table I*).

Seven (70%) neonates had no associated congenital anomaly, while 3 cases (30%) had multiple congenital anomalies (*Table II*), in 2 of which, they were incompatible with life.

Grossly, the liver, spleen and kidneys were mild to moderately congested at autopsy, in 8 cases (80%), while in 1 case, the left lobe of liver showed compression atrophy.

Microscopically, the lungs showed evidence of unilateral left-sided hypoplasia in 4 cases (40%) (*Fig. 2*); bilateral congenital atelectasis 7 cases (70%); pulmonary hemorrhage, edema and congestion –5 cases (50%) and extramedullary hematopoiesis–3 cases (30%).

Discussion

The incidence of congenital diaphragmatic hernia varies from 1 in 2000 to 1 in 5000 livebirths(1,2). An incidence of 1.4% of all necropsies; 0.95% of necropsied stillbirths and 1.9% of necropsied neonatal deaths, has been reported(3). In the present study, of a total of 588 autopsies, 10 cases of congenital diaphragmatic hernia were encountered *i.e.,* an incidence of 1.7% of perinatal necropsies. It is difficult to give an accurate figure for the incidence, because some infants die undiagnosed in early neonatal life without necropsy; in addition some of them do not present until late childhood or adult life.

Barring male predominance, no racial or regional preference has been noted and most maternal factors showed no consistent relationship. However, hydramnios has been noted in 20% of pregnancies

TABLE I—*Clinical Details of Cases of Congenital Diaphragmatic Hernia*

Infant characteristics		No. of cases (n=10)	Percentage
Viability	Liveborns	8	80
	Stillborns	2	20
Age at death (h)	0 to 8	7	70
	8 to 24	—	—
	>24	1	10
Sex	Male	7	70
	Female	3	30
Weight at birth (kg)	<2.0	8	80
	>2.0	2	20
Nature of delivery	Normal vaginal	7	70
	Forceps vaginal	2	20
	Cesarean section	1	10
Accurate clinical diagnosis of congenital diaphragmatic hernia at birth		6	60
Left-sided posterolateral diaphragmatic defects		10	100
Herniated contents	Intestinal loops	8	80
	Left lobe of liver	5	50
	Stomach	4	40
	Spleen	3	30

Clinical features of liveborns with congenital diaphragmatic hernia	No. of cases (n=8)	Percentage
Dyspnea & cyanosis within 6 min of birth	5	62.5
Dextrocardia with displaced apex beat	8	100.0
Scaphoid abdomen	3	37.5
Decreased breath sounds	5	62.5
Increased percussion dullness	3	37.5
Bowel sounds in chest	3	37.5

Maternal characteristics		No. of cases (n=10)	Percentage
Parity	Multigravida	6	60
	Primigravida	4	40
Period of gestation	Fullterm	7	70
	Preterm	3	30
Complication in pregnancy	Eclampsia	1	10
	Uncomplicated	9	90

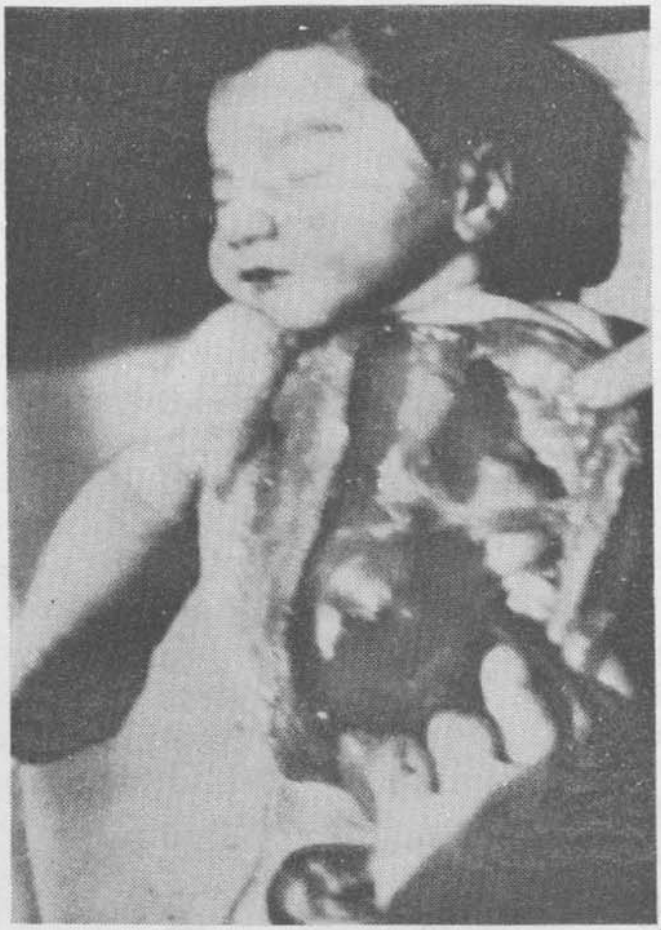

Fig. 1. Autopsy photograph of an infant with left-sided posterolateral diaphragmatic defect showing part of the stomach lying within the thoracic cavity.

resulting in the birth of a child with congenital diaphragmatic hernia(3). In the present study, a male:female ratio of 7:3 was observed. Maternal factors were inconsistent and in no case maternal hydramnios was detected.

In newborns with the common left-sided defect: cyanosis, dyspnea and dextrocardia have been reported(4). In the present study, this classical triad was seen in 50% cases. *X*-ray chest has been reported to be diagnostic, revealing mediastinal shift, air-filled bowel loops in the chest and a relatively gasless abdomen(4). In the present series, 8 cases showed the presence of intestinal loops within the thoracic cavity at autopsy.

An accurate clinical diagnosis of congenital diaphragmatic hernia at birth was possible in only 6 of the 10 cases (60%). However, the presence of extremely low Apgar scores immediately at birth, in most cases, particularly when initial investigations were not possible and clinical examination was the mainstay of the diagnosis, obviously rendered an

Fig. 2. Microphotograph of hypolastic lungs showing immature bronchi, widened alveolar septae, ill-formed nonaerated alveoli and primitive mesenchyme (H&E × 40)

TABLE II—*Associated Congenital Anomalies in Cases of Congenital Diaphragmatic Hernia*

No anomaly		7
Multiple congenital anomalies		3
Case 1	—	Cervical lymphangioma Congenital splenomegaly Adrenal rest in testis
Case 2	—	Congenital absence of cervical vertebrae Atrophy brain Hydrocephalus Congenital bilateral talipes equino varus Aorta arising from right ventricle, rudimentary pulmonary trunk showing only one semilunar valve.
Case 3	—	Accessory lobe of liver Conjoint adrenals Meningomyelocele Meckel's diverticulum

accurate clinical assessment more difficult.

Left-sided posterolateral diaphragmatic defects have been noted to occur 8 times more frequently than right-sided hernias in newborns, while bilateral hernias are rare (1%), and usually fatal(5). In the present analysis, left-sided posterolateral defects were seen in all 10 cases studied, while no bilateral hernia was seen. The commonest herniated contents were the intestinal loops (80%) and the left lobe of liver (50%).

Excluding malrotation and patent ductus arteriosus, anomalies related to neural axis, skull, vertebral column have been reported with an incidence between 10-20% of liveborns(6,7). This rises to 95% in stillborns(3). In our study, congenital anomalies of the neural axis, skull, vertebral column and cardiovascular system were seen in one of the 8 liveborns (12.5%) and in both the stillborns (100%) and were major factors contributing to mortality.

In congenital diaphragmatic hernia, pulmonary hypoplasia is reported to be associated with decreased size and number of bronchi, lung saccules and alveoli. There is also a corresponding decrease in the total size of the pulmonary vascular bed(8,9). In the present series, 4 neonates suffered from associated severe unilateral left-sided pulmonary hypoplasia and the extent of this hypoplasia was a major contributory factor to respiratory failure and death. Two criteria were used in evaluating pulmonary maturity: first, the comparative weight of the right and left lungs, as compared to the expected weight for age of newborns(10,11); second, the macroscopic and the microscopic findings, such as the degree of alveolar and bronchial immaturity(12).

Notwithstanding surgical intervention, infants with severe respiratory distress and cyanosis within the first 24 hours of life, have a 50% mortality, partly due to associated congenital abnormalities(13). In the present study, 7 neonates died within 8 hours of birth, while 1 case died at 37 hours, and 2 were stillborn.

It is suggested that the possibility of a congenital diaphragmatic hernia should be considered in any newborn who developed severe respiratory distress, cyanosis with dextrocardia and has an indefinable diaphragm and a cystic gas-filled lesion on chest *X*-ray. In recent years, the availability of ultrasound, amniography and computed tomography and the consideration of immediate fetal surgery have directed attention to the antenatal diagnosis of congenital diaphragmatic hernia. The proposed advantage of antenatal diagnosis is the ability to deliver the infant electively at a medical centre prepared to care for the complex problems of the infant as well as those of the mother. Surgical success has been achieved, but we recognize that the barrier to survival is pulmonary parenchymal and vascular hypoplasia as well as the complex syndrome of persistent fetal circulation.

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