

## Original Article

# Congenital diaphragmatic hernia: A fetal study

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### ABSTRACT

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**Background:** Congenital diaphragmatic hernia (CDH) is one of the rare types of malformations in the new born. The estimated incidences of CDH is 1 in 2000 to 5000 live births.

**Methods:** This study was conducted on 1000 fetuses obtained from the Department of Obstetrics and Gynecology, Government Medical College and Hospital, Chandigarh. The autopsies were done in the Department of Anatomy, GMCH Chandigarh.

**Results:** In our present study, the incidence of CDH was 1.6 % (out of 1000 fetuses, 16 fetuses had CDH) among all the cases. The left-sided defect was seen in 75% of the cases, right sided in 12.5% of cases and bilateral and central tendon defect in 1% of the cases. Associated anomalies were noted in each case.

**Conclusion:** Prenatal diagnosis of CDH is important to detect and avoid further complications in life.

**KEYWORDS:** Congenital diaphragmatic hernia; pulmonary hypoplasia; herniation.

## Introduction

Diaphragm is the musculo-fibrous partition between the thorax and the abdominal cavity. Defect in the diaphragm may cause herniation of structures of the abdominal cavity into the thoracic cavity. This defect may be congenital or acquired.

Congenital Diaphragmatic Hernia (CDH) permits the herniation of abdominal contents into the thorax through an orifice in the diaphragm; it most commonly occurs on the posterolateral aspect on the left side.<sup>1</sup>

According to Torf et al, the prevalence of defects in the diaphragm among newborns is 1 in 3000.<sup>2</sup> Left-sided posterolateral hernias are more common (85%) than right-sided (15%) because the liver plugs the defect whereas central tendon defects and bilateral CDH are very rare (2 to 3%). The most common anomaly associated with CDH is pulmonary hypoplasia which is due to pressure effects from herniated structures such as the liver, stomach, spleen and intestine.<sup>4</sup> This leads to poor development

of the terminal bronchioles, alveoli and pulmonary vessels resulting in respiratory failure soon after birth.<sup>5,6</sup> According to Sweed et al, other congenital anomalies associated with CDH accounts for about 30 to 40% of cases, which include neural tube and cardiac defects, chromosomal and craniofacial anomalies, omphalocele and urinary tract and gastrointestinal defects.<sup>7</sup> This study evaluated prevalence of congenital diaphragmatic hernia in aborted fetuses.

## Materials and Methods

This study was conducted on 1000 fetuses obtained from the Department of Obstetrics and Gynecology of the Government Medical College and Hospital, Chandigarh from 2008 to 2015. These fetuses were obtained as a result of spontaneous abortions and IUDs. In some cases, MTP was indicated after the detection of defect on antenatal ultrasound. Out of 1000 fetuses, 16 were found to have congenital diaphragmatic hernia. The family history and medical and occupational history of parents was noted. Obstetric and antenatal history of each mother was noted. Each fetus was examined externally. Photographs and radiology of each fetus with the defect was also done. Autopsy was performed following the routine procedure. The internal examination was done and other organs/systems with any anomaly were also noted and correlated with the history.

## Observations

Out of 1000 fetuses, 16 (1.6%) were found to have congenital diaphragmatic hernia. The type of defect and its frequency are as shown below .

| Type of defect                                   | No of cases (%) |
|--|-----------------|
| Left sided Bochdalek hernia ( <b>Figure 1</b> )  | 12 (75%)        |
| Right sided Bochdalek hernia ( <b>Figure 2</b> ) | 2 (12.5%)       |
| Bilateral hernia ( <b>Figure 3</b> )             | 1 (6.25%)       |
| Central tendon defect                            | 1 (6.25%)       |

| Associated anomalies | No of cases (%) |
|----------------------|-----------------|
| Cardiovascular       | 4 (25%)         |
| Respiratory          | 9 (56%)         |
| Neural tube defects  | 10 (62.5%)      |
| Gastrointestinal     | 8 (5%)          |
| Urogenital           | 2 (12.5%)       |
| Musculoskeletal      | 10 (62.5%)      |
| SUA                  | 2 (12.5%)       |

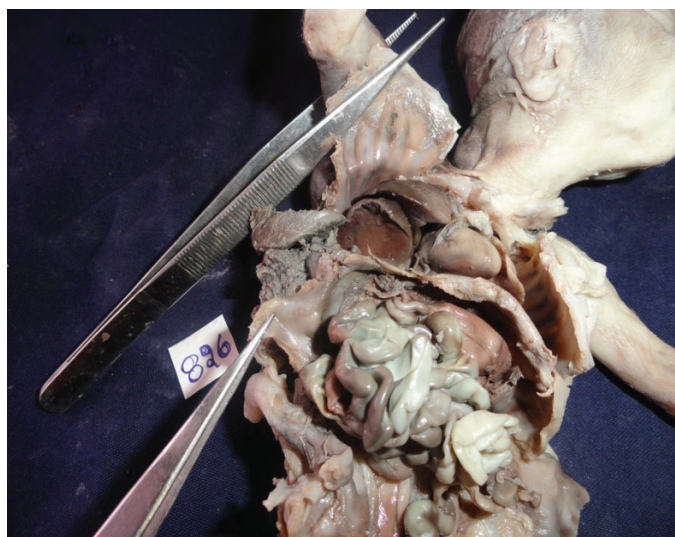
| Herniated structures | No of cases (%) |
|----------------------|-----------------|
| Liver                | 15 (93%)        |
| Gall bladder         | 1 (6.25%)       |
| Stomach              | 12 (75%)        |
| Spleen               | 11 (68.7%)      |
| Small intestine      | 7 (43.7%)       |
| Appendix             | 1 (6.25%)       |
| Kidney               | 1 (6.25%)       |



**Figure 1: Left sided diaphragmatic hernia.**

## Discussion

The diaphragm is a musculo-fibrous partition that separates the thorax from the abdominal cavity. It consists of two cupolae and a central tendon.<sup>8</sup> Development of the diaphragm starts by the third week and is completed by the eighth week of intrauterine life.<sup>9</sup>



**Figure 2: Right sided diaphragmatic hernia with lateral body wall syndrome.**

CDH is one of the most common malformations in the new born and is commonly caused by failure of closure of one or both pleuro-peritoneal canals.<sup>10</sup> There are four types of congenital diaphragmatic hernia: (a) Hiatal hernia : herniation through a congenitally large esophageal orifice (3%) (b) Bochdalek Hernia (posterolateral defect): this is the most common type of congenital hernia and is seen mostly in males. Incidence of bilateral hernia is 5%, while unilateral hernia is seen in about 90% of cases. (c) Morgagnian hernia: the defect is in the anterolateral part of the diaphragm. This defect has a female preponderance. It accounts for 2% of all the cases. (d) Defects occurring in the central tendon: It is the rarest type of diaphragmatic hernia reported.<sup>11</sup>

In 1960, Lazarius reported the first case when he found the defect accidentally during autopsy.<sup>12</sup> In a study conducted on CDH in Australia, it was observed that about 70 to 75% of cases had posterolateral defects, 23 to 28% Morgagnian hernia and only 2 to 7% were with central defect.<sup>13</sup> Another study by Benjamin et al mentioned that left-sided posterolateral hernia was more common (85%) than right-sided (13%). The incidence of bilateral hernia was 2%. They also noted a male preponderance in left-sided hernia with a male to female ratio of 3:214 whereas the right sided hernia had a male to female ratio of 3:111. A retrospective study of neonatal autopsies done in New



**Figure 3: Bilateral congenital diaphragmatic hernia.**

Delhi during the span of 30 years encountered 10 cases of CDH among 588 autopsies done. They concluded that CDH accounted for about 1.7% of all cases. They also noted that there was no relation between any possible maternal factors and CDH. Male female ratio was 7:3. The left lobe of the liver and intestinal loops were the common herniating structures.<sup>15</sup>

Studies have mentioned that left-sided Bochdalek hernia are more common than right-sided. Levy et al reported that the frequency of left-sided posterolateral diaphragmatic defects was 8 times more than right-sided hernias in newborns.<sup>16</sup> In our study, we noted 14 cases of unilateral CDH out of which 12 cases were left-sided. The left side of the diaphragm closes later which may be the reason why the CDH is seen more on left side.<sup>17</sup> We noted a case of bilateral CDH with herniation of the liver, coils of the intestine, the stomach and the spleen. Less than 1% of neonates had bilateral CDH as reported.<sup>18</sup>

Right-sided posterolateral hernia is rare compared to left-sided. A protective effect of the liver developing in the septum transversum and early closure of the pleuroperitoneal opening can explain why right-sided

CDH occurs early<sup>19</sup> although apart from this, the presence of a right-sided CDH could also be missed because of similar echogenicity of herniated liver with that of the lung.<sup>20</sup> Two cases of right-sided CDH with herniation of the liver and gall bladder were part of our study group.

The diaphragmatic defects can be detected as early as in the second trimester by ultrasonography.<sup>21</sup> Three cases of CDH in our study were diagnosed ultrasonographically. The rest of the cases were detected during autopsy. By finding out direct signs like the presence of abdominal organs in the thorax and indirect signs like mediastinal shifts, abnormal cardiac axis and the presence of polyhydramnios on ultrasound, a diagnosis of CDH can be made. Van den Hout L described a case series where a prenatal diagnosis of CDH was missed in 40 per cent of cases and these infants postnatally showed acute respiratory distress, absence of breath sounds on the ipsilateral side, shifted cardiac sounds and bowel sounds in the chest, a barrel-shaped chest and a scaphoid abdomen.<sup>22</sup> Jain et al showed that herniation of the liver detected by radiological investigations may be associated with a poor prognosis.<sup>9</sup>

## Conclusion

Clinicians should be careful regarding bilateral CDH even though it is an extremely rare congenital malformation since this is one congenital anomaly which can be detected and treated prenatally to reduce mortality and postnatal outcomes. Genetic factors play an important role in the development of CDH. Parents should be informed about the severity of CDH, the expected pre- and post-natal events and the risk of poor outcomes including death and the various long-term morbidities.<sup>23</sup>

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