Case Report

Bilateral Huge Congenital Diaphragmatic Hernia in a Neonate with Three Liver Lobes: A Case Report

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ABSTRACT

Bilateral Congenital Diaphragmatic Hernia (CDH) is a birth defect with a rare occurrence. The neonates who are born with this defect are not able to survive to undergo the treatment. In this case report, we provide the details of anatomical abnormalities and changes in organ development observed in a male neonate with a huge diaphragmatic defect which was undiagnosed till after birth.

ABBREVIATIONS

PAPP-A: Pregnancy Associated Plasma Protein A, AB: Antibody, AFI: Amniotic Fluid Index, AFP: Alfa Feto Protein, CDH: Congenital Diaphragmatic Hernia, HBs Ag: Hepatitis B Surface Antigen, HCV: Hepatitis C Virus, HIV: Human Immune Deficiency Virus, TFT: Thyroid Function Test

INTRODUCTION

Congenital diaphragmatic hernia (CDH) is a major birth defect, in which some parts of the abdominal contents protrude into the chest cavity [1]. Its occurrence is about 1 in 2000-3000 births [2]. An extremely rare variation of CDH is the bilateral form that accounts for approximately 1% of all CDH patients [3-5]. Here, we report a case of bilateral huge congenital diaphragmatic hernia, not diagnosed during prenatal assessments.

CASE REPORT

A male baby was born from a 27-year-old primigravida mother at gestational age of 37 weeks and 5 days by normal vaginal delivery in a private hospital. The labor pain was started upon spontaneous rupture of membrane about 5 hours before delivery at home with clear fluid. Delivery stages moved smoothly without any problem. At delivery, the neonate appeared cyanotic, and apnoeic. The heart rate was initially over 100 beats/min with undetectable blood pressure. Cord blood gas was pH 7.24, PCO2 52.7 mmHg, PaO2 18.5 mmHg, HCO3 22.6 mmol/L and calculated base deficit was 5.6 mmol/L. The baby had APGAR scores of 2 at one and five minutes and 1 at ten minutes without any positive response to the cardiopulmonary resuscitation which was done based on the standard neonatal resuscitation protocol [6]. The baby was intubated and transferred to Neonatal Intensive Care Unit (NICU) and kept under mechanical ventilation support with positive pressure. Chest X- Ray (CXR) including abdomen showed presence of hypoaerated small lungs, solid organs in the bilateral chest cavity extending to the upper sides, displaced heart and thymus shadow to the right, an empty organ (possibly the

stomach) and some small air-filled areas in between them (Figure 1a). Bowel loops were visible in the abdomen.

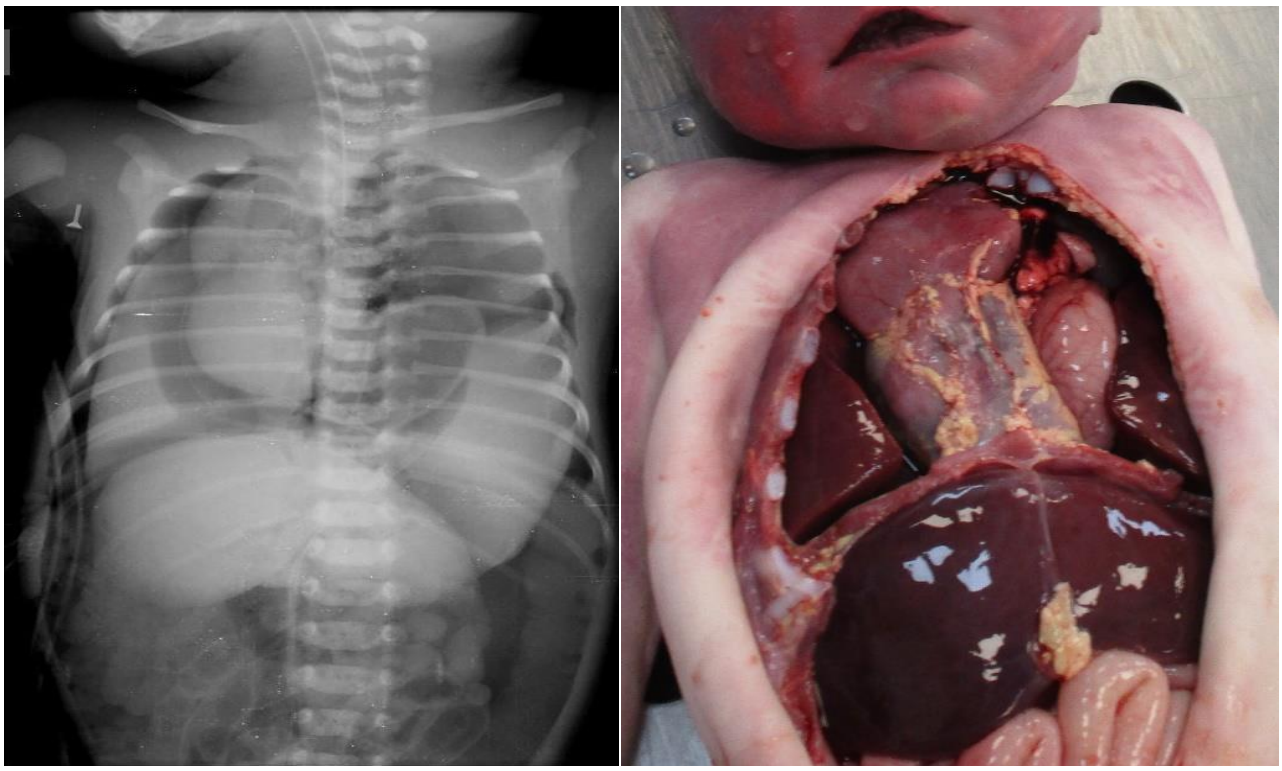

Figure 1: Comparative view of a) CXR and b) Autopsy of the patient.

a) CXR:
Presence of abdominal solid organs and stomach in the thoracic cavity

b) Autopsy view
Presence of three liver lobes; two lobes in the thorax and one in the epigastric region. Stomach is neighbouring the heart.

Due to the unresponsiveness to CPR and small air-filled areas observed in the CXR, chest tube was inserted with draining small amount of air. CXR after insertion showed proper removal of that small amount of pneumothorax. Unfortunately, despite of high-pressure ventilator support, maintaining intravenous fluid, administration of epinephrine, dopamine and cardiac compression the neonate expired about 1 hr after birth. The pregnancy and delivery were uncomplicated. There was not any clear history of smoking, alcohol consumption or usage of any medication except the perinatal vitamins. The parents were cousin and healthy without any family history of fetal or neonatal death. All of the perinatal blood screening tests in the pregnancy including HIV, HBs Ag, HCV, AFP, Anti rubella Ab, TFT (TSH, T4, T3), PAPP-A, and screening for trisomies 18 and 21 were reported as normal. Ultrasounds at 12 and 19 weeks revealed normal appearance of fetal brain, spine, spinal cord, kidneys, bladder, stomach and heart.

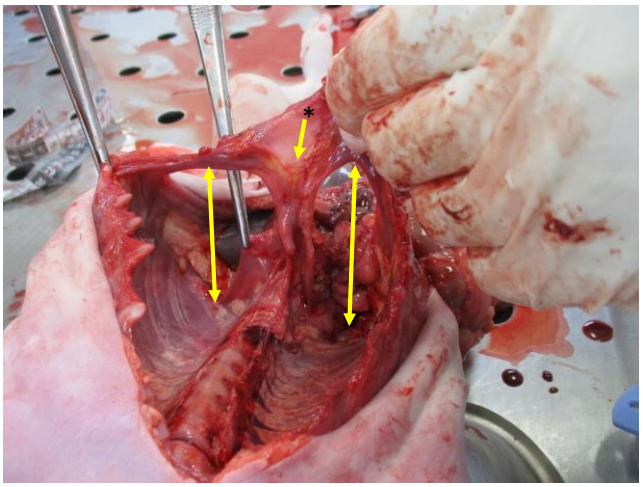

Figure 2: Bilateral huge diaphragmatic defect. The arrows show the defects in both sides. Abdominal viscera are seen at the back of the arrows. * The arrow in the middle indicates the partially developed central tendon.

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Due to normal heart structure and function observed in the fetal sonographies, echocardiography was not requested. However, polyhydramnios was reported at 34 weeks and 5 days (AFI: 23-24 cm), which was his last prenatal sonographic evaluation. On physical examination, the infant's abdomen was not scaphoid, and musculo-skeletal system, palmar creases, nails and face appeared normal. His birth weight was 2825 gram (percentile 50), the head circumference was 33 cm (percentile 50) and height 49 cm (between 50-90 percentiles), all normal range. Autopsy (after parental consent) was done in Shiraz Department of Forensic Medicine. The general view of chest cavity is shown in Figure 1, b. As can be seen, it is in accordance to the CXR showing the presence of solid organs in bilateral hemithoraces as well as the stomach in the left hemithorax. Further evaluation revealed bilateral huge diaphragmatic defect with a small muscular rim. The defect was measured about 6 cm in each hemidiaphragm (approximately 90% of each side) (Figure 2).

A thin remnant of central tendon of diaphragm extended from the anterior chest wall to the spine separating two sides of the defect. The lungs were extremely hypoplastic; left lung was 3 cm and the right one was 2.5 cm with the weight of 5 grams for each. The majority of the pleural cavity was occupied with the liver (15 × 8 cm, 115 gr), stomach, spleen, and duodenum. The liver had three lobes; the middle one including gallbladder remained in the abdominal cavity while the right and the left lobes were shifted to the thorax. Thymus was normal for the gestational age (15 grams). The heart was normal with four chambers (weighed 15 gr). Aorta, its arc and pulmonary artery were all in the normal positions. In abdominopelvic cavity, small and large intestine, kidneys, pancreas and bladder were normally seen without malrotation. Right and left adrenal glands appeared normally; the left one weighed out 5 gr which was upper limit of its size for the gestational age [7]. He was phenotypically male with normal penis and testicles. Unfortunately, chromosomal study was not performed.

DISCUSSION

Diaphragm develops through a complex process in which 4 independently developing structures join together: the pleuroperitoneal membranes, the septum transversum, the dorsal mesentery of the esophagus, and the lateral body walls [8]. These structures normally fuse together during weeks 4–8 of embryologic development. In brief, within week 7, pleuropericardial membranes join together to build the pericardium. This separates the pericardial cavity from the pleural cavities. Pericardial and peritoneal cavities are divided by central tendon which is made mainly by the contribution of septum transversum. Peritoneal and pleural cavities are separated from each other by development of pleuroperitoneal membranes. This completes the compartmentalization. Any developmental failure of these membranes results in diaphragmatic hernia [9]. Based on the anatomical definition, there are four types of CDH [1]: (a) Hiatal hernia: herniation through a congenitally large esophageal orifice, (b) Bochdalek Hernia: posterolateral defect, the most common type and mostly in males, (c) Morgagnian hernia: anterolateral defect in diaphragm and female preponderant, (d) Central tendon hernia: the rarest type [1]. In our case, the anomaly represents defective formation of the pleuroperitoneal membrane, and some parts of septum transversum with partial development of a diaphragmatic remnant from the lateral body walls. As this major defect occurs in early stages of the pregnancy, liver and spleen grow in the pleural space, and consequently, the lungs become hypoplastic.

There are indirect sonographic findings that should provide clues to search for CDH: polyhydramnios, cardio-mediastinal shift, cardiac axis abnormality and absence of the normal stomach bubble [10]. Generally, the diagnosis of CDH can be certain from the true transverse plane study with following findings: absent bowel loops in the abdomen, intrathoracic herniation of the liver (presents in up to 85% of cases and with a worse prognosis but it is difficult to be diagnosed due to the same echogenicity of liver with its neighbouring lung tissue), peristaltic bowel movements in the chest and decreased abdominal circumference due to upshifting of organs to the thorax [11].

There are some sonographic specifications to differentiate the left-sided from the right sided CDH. In the left-sided CDH, the findings are as follows [11]: the heart on four- chamber view is at the same transverse level as the stomach and small bowel and superior to the inferior margin of the scapula. Moreover, the gallbladder is displaced to the left. In the right-sided CDH, the sonographic clues are described as following: the umbilical

segment of the portal vein bows leftward. Using color doppler study, the portal branches bow to the lateral segment of the left hepatic lobe and passing towards or above the diaphragm. The gallbladder displaced above the diaphragm. Furthermore, the left hepatic lobe appears as an echogenic space between the left heart border and stomach [10]. In addition to the mentioned anatomical displacements, the degree of pulmonary hypoplasia can be calculated and correlates with the observed-to-expected lung-to-head ratio (O/E LHR) with sonography or more accurately with MRI [12,13].

In our case, polyhydramnios was the only reported sonographic clue. Thus, there was no suspicion to search for CDH. Based on the previous studies, the echogenicity of the liver is very similar to that of the lung. Therefore, it can be difficult to assess the liver position or its extent of displacement into the thorax [14]. However, diaphragmatic defects can be visualized through sagittal scanning through the fetal body. Using color Doppler ultrasound examination, ductus venosus and intrahepatic vessels in the thoracic cavity are identified. In addition, MRI is more accurate in demonstrating the abnormal position of the fetal liver compared to ultrasonography [12,15]. Besides, Cardiovascular defects are the most common anomalies associated with CDH [16]. The heart was normal four chambers in our case with normal cardio mediastinal axis since the abdominal solid organs moved up symmetrically. In addition, abdominal circumference was compatible with the gestational age because bowel loops remained in the abdominal space, and also no bowel loop sign was observed in the thorax. Furthermore, CDH may accompany with bilateral renal hypertrophy, omphalocele, skeletal abnormalities such as spina bifida and sternum bifida, genitorynary malfomations, etc. [17] which our case did not have any of them in antenatal sonographic evaluation. Besides, the stomach was placed in the left lower hemithorax in CXR of our case but there was no fetal sonographic report of its thoracic position. “The stomach bubble may not be visualized due to its transient emptying at the time of sonography. Therefore, it is not significant in the absence of the other prenatal findings” for CDH [18]. Although this bilateral huge CDH could be diagnosed by further prenatal imaging evaluation, this was not done due to lack of possible clues in his routine sonographic evaluation. It would be better if the portal vein and its branches were carefully examined in this case during serial prenatal sonographic follow ups. So, the latter is to be included in the routine sonographic check-up rather than suspected cases. Evaluation of the location of the gallbladder could not be helpful in the prenatal diagnosis of CDH in such cases as it stayed in the abdominal cavity together with the middle lobe of the liver.

Since in the perinatal and family history, the neonatal physical examination and autopsy there was not any anomalous observation, our case was considered a sporadic isolated CDH. This is typically suspected or diagnosed during mid-gestation ultrasound evaluation. Then, the patient will be reassessed with targeted ultrasound and Magnetic Resonance Imaging (MRI) to confirm the diagnosis. Fetal therapy, such as fetal Fetoscopic Endoluminal Tracheal Occlusion (FETO), can be offered to the patients with predicted poor prognosis. It is one of the most effective procedures to trigger intrauterine lung growth. It is performed within 27-32 weeks of gestation depends on the severity of CDH [15]. In addition, the occurrence of persistent pulmonary hypertension may be reduced by administration of prenatal transplacental sildenafil, either alone or in combination with fetal surgery [19]. Furthermore, Zaupa et al. in 2007 reported a case of bilateral CDH with gastroschisis with a favorable outcome [20] which shows there might be potentials for development of some techniques in the future to reduce fetal intra-abdominal pressure to further promote intra-uterine growth of lungs.

CONCLUSION

This was a rare case of bilateral huge congenital diaphragmatic hernia with poor outcome. Despite of non-favourable outcomes for bilateral CHD patients, early prenatal diagnosis is very important. Because, it provides opportunities for using the potential intra-uterine operation techniques to correct the defect, and also, giving proper counselling regarding the pregnancy outcome.

DECLARATION OF INTERESTS

The authors report no conflict of interest on this paper.

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