Ultrasonic Diagnosis of Congenital Diaphragm Hernia
And Successful Outcome After Postnatal Surgery

Yusrawati¹, Putri Mentari Faisal²

¹Division of Maternal-Fetal Medicine Department of Obstetrics and Gynecology, RSUP Dr. M. Djamil, Andalas University, Padang, Indonesia
²Specialist Doctor Education Program (PPDS) in Obstetrics and Gynecology, Faculty of Medicine, Andalas University, Dr. M. Djamil Padang

ABSTRACT

Background: Congenital diaphragmatic hernia (CDH) is a disorder where abdominal contents protrude into the chest. Congenital diaphragmatic hernias are seen in 1 of every 2000-4000 live births. Mortality is predominantly due to the development of pulmonary hypoplasia which is thought to be due to the mass effect on the developing lung.

Case Report:Mrs. Y, a 32-year-old controlled for antenatal care at the fetomaternal polyclinic M Djamil hospital. She had no complaints, the patient was referred from another hospital with a suspected fetal diaphragm hernia. The patient is known to be 38–39 weeks term pregnancy, from ultrasound examination found herniation of abdominal contents into the thoracic cavity and the liquor was excessive and the doppler study was normal. The ultrasound showed normal growth and corresponded to 38 week Then the patient was planned for caesarean section and a hernia repair surgery. The neonate was admitted to the neonatal intensive care unit (NICU). He was shifted to the pediatric surgeon with ventilator support after 48 h and was undergoing surgery. There was a posterolateral left sided defect (BOCKDALEK type) of around 22–24 mm. The intestines were reduced to the abdomen and the closure was done. Immediately after the surgery, the child was admitted back to the NICU and the further postnatal course was uneventful.

Discussion and Conclusion: CDH is a malformation of the diaphragm due to failure of closure of the pleuroperitoneal canal at around 10 weeks leading to herniation of abdominal contents into the thoracic cavity leading to compression of lung tissues at pseudo glandular stage. CDH is often ultrasound diagnosed before birth [120, 121]. The intestine and/or the liver may be in the thorax and the lungs are small. US scan allows detailed assessment of the heart. The survival depends on the contralateral lung volume, Ninety percent of cases are due to posterolateral defect which is also called Bochdalek hernia—80% are left sided, 15% are right sided and 5% are bilateral. Herniated viscera leads to pulmonary hypoplasia and pulmonary hypertension. Prognosis depends on the gestational age at diagnosis (earlier the diagnosis, likely larger the defect), associated anomalies, contralateral lung volume, pulmonary vasculature (Doppler), cardiac function, liver herniation and gestational age at delivery.

KEYWORDS
Congenital diaphragmatic hernia, prenatal diagnosis, prenatal ultrasound diagnosis

CORRESPONDENCE
Phone: 081285483420
E-mail: Mentari.faisalputri@gmail.com
I. INTRODUCTION

Congenital diaphragmatic hernia (CDH) is a developmental disorder that occurs in 1-4 cases per 10,000 births. In this disorder there is a defect in the diaphragm that allows the abdominal organs to herniate into the thoracic space through the diaphragm. In development the diaphragm is formed during the 4th and 12th week of pregnancy. A defective hole in the diaphragm can occur when the pleuroperitoneal membrane fails to close at the end of organogenesis. When the fetal midgut migrates back into the abdominal cavity (usually by week 10-12) pressure forces the abdominal organs to herniate through the defect in the diaphragm.

The most common type of defect is the pleuroperitoneal membrane closure defect known as a Bochdaleck hernia accounting for 90% of all CDHs. It usually occurs as a posterolateral defect affecting the left hemithorax in the majority of cases. The clinical presentation depends on the extent of the pulmonary hypoplasia and can vary from cyanosis and respiratory distress immediately after birth to the final presentation of respiratory distress in the first few days of life when the herniated bowel loop fills with air and compresses the ipsilateral lung parenchyma. Typical signs include abnormal heart impulses, absent breath sounds, and a scaphoid abdomen.

The diagnosis is usually made by prenatal ultrasonography and confirmed on a postnatal chest radiograph.

Complications to watch for in neonates with CDH include pulmonary hypoplasia and primary pulmonary hypertension. The mass effect of the herniated organ to the chest does not allow the lungs to expand while the fetus is developing. Neonates born with this are usually placed on extracorporeal membrane oxygenation (ECMO) to displace oxygen if the neonate's lungs are hypoplastic. In more complicated cases, 15% to 45% of fetuses also present with other abnormalities and anomalies. These include heart defects and chromosomal defects such as trisomy.

The prognosis in cases of CDH is associated with several risk factors that can be assessed prenatally, including hepatic herniation, gastric herniation, low fetal lung volume, and low head-to-lung ratio (LHR) on prenatal ultrasound, as well as the presence of chromosomal abnormalities, concomitant anomalies, and hydrops fetalis.

II. METHODS

Case report according to our case report, that can be helpful in term of providing neonatologist, surgeon and maternal-fetal medicine specialist with realistic prognostic information for counseling families and to diagnose the CDH. Furthermore, it was recently established a multidisciplinary CDH clinic that was composed by specialists from areas, including pediatric surgery, pulmonary, cardiologi and nutrition.

III. RESULT

We investigated a patient Mrs. Y, a 32-year-old that controlled for antenatal care at the fetomaternal polyclinic m djamil hospital. She had no complaints, the patient was referred from another hospital with a suspected fetal diaphragm hernia. Physical Examination On physical examination, vital signs were within normal limits, with a normal female body appearance. The patient's BMI of 23.4. Head and neck examination did not reveal webbed neck or facial malformations. Physical examination of the chest, lung, and heart was also normal. On abdominal examination, there was tenderness and a palpable mass in the upper right region. Vaginal toucher didn't perform. The patient is known to be 38-39 weeks term pregnancy, from ultrasound examination found herniation of abdominal contents into the thoracic cavity and, the liquor was excessive and the doppler study was normal. The ultrasound showed normal growth and corresponded to 38 weeks.
Then the patient was planned for caesarean section and hernia repair surgery. The neonate was admitted to the neonatal intensive care unit (NICU). He was shifted to the pediatric surgeon with ventilator support after 48 h and was undergoing surgery. There was a posterolateral left sided defect (BOCKDALEK type) of around 22–24 mm. The intestines were reduced to the abdomen and the closure was done. Immediately after the surgery, the child was admitted back to the NICU and the further postnatal course was uneventful.
IV. DISCUSSION

Congenital diaphragmatic hernia (CDH) is defined as a rare developmental disorder of diaphragm formation, characterized by herniation of the abdominal organs into the chest cavity which can then lead to varying degrees of pulmonary hypoplasia and pulmonary hypertension. These abnormalities can vary in size and include agenesis of the diaphragm, well-defined pinholes, and less commonly, thinning or undermuscularization of the diaphragm tissue. The incidence of CDH based on available literature ranges from 0.8-5/10,000 births and varies across populations. Based on the literature notes, it was found that there was a slightly higher male predominance and a lower risk of isolated CDH was reported among African Americans. Although patient survival has increased with the advancement of advanced diagnostic techniques along with medical and surgical treatment, the worldwide average mortality rate is 50%. In patients there may be long-term morbidity, duration of postnatal hospital stay of affected neonates, poor growth, developmental delay, gastroesophageal reflux, and chronic oxygen dependence. The etiology of CDH remains largely unclear and is currently considered to be multifactorial. Most cases have isolated diaphragmatic defects with pulmonary hypoplasia and persistent pulmonary hypertension of the newborn (PPHN). CDH may be associated with cardiac, gastrointestinal, genitourinary anomalies or with chromosomal aneuploidies such as trisomy.
abnormalities often involve the cardiovascular, central nervous, and/or musculoskeletal systems. Cardiovascular malformations (ventricular septal defect, atrial septal defect, tetralogy of Fallot) occur in 11%-15% of isolated CDH cases and 25%-40% of all CDH cases. Central nervous system disorders such as neural tube defects and hydrocephalus occur in 5%-10% of cases of nonsyndromic CDH. Limb anomalies such as polydactyly, syndactyly, and limb reduction defects occur in approximately 10% of cases of nonsyndromic CDH. Chromosomal abnormalities including aneuploidy, chromosomal deletions/duplications, and complex chromosomal rearrangements are identified in 10%-35% of CDH cases who are not isolated and diagnosed before birth. Trisomy 13, 18, 21 and 45, X are the most common aneuploidies associated with CDH.6

There is growing evidence that environmental factors and specific pathways play a role in the development of CDH. Studies in rodent models show that interference with the vitamin A pathway plays a role in CDH. This was observed when 25–40% of offspring of mother rats fed a diet deficient in vitamin A developed CDH, and the proportion of affected pups decreased when vitamin A was reintroduced to the diet in mid-gestation. Two clinical studies have shown that plasma retinol and cord blood levels of retinol-binding protein were significantly lower in newborns with CDH compared with controls, irrespective of maternal retinol levels. In addition, exposure to nitrofen, herbicides in rodents has also been shown to cause CDH in most of their offspring.6,8

The embryological basis of CDH is still controversial. It was originally thought that the defect was secondary to failure of various parts of the diaphragm to fuse resulting in a patent pleuroperitoneal canal. This can then allow the intestine to enter the thoracic cavity when it returns from the umbilical extraembryonic coelom. Another speculation is that pulmonary hypoplasia may be a major causal factor in the pathophysiology of diaphragmatic hernia. If the development of the lung buds is disturbed, there is a disturbance in the development of the post hepatic mesenchymal plate (PHMP) which is closely related to lung development, resulting in a diaphragm defect.8 Regardless of the basis, defects in the diaphragm cause the abdominal organs to herniate into the thoracic cavity resulting in abnormal lung development. This defect also causes abnormal fetal breathing movements that result in vacuous maturation of the lungs due to stretching. Lung changes include decreased terminal bronchiolar branching leading to acinar hypoplasia with fewer alveoli, reduced areas of gas exchange, thickened alveolar walls, and increased interstitial tissue. Thus the main pathophysiology underlying CDH appears to be a combination of pulmonary immaturity and hypoplasia leading to persistent pulmonary hypertension of the newborn (PPHN).6,8

Lung hypoplasia occurs on the ipsilateral side of the herniation, with the contralateral side affected to varying degrees. Hypoplasia was originally thought to be secondary to physical compression of the lung by stomach contents that restrained lung expansion. Recently, a “two-hit” hypothesis has been proposed based on a mouse model explaining lung injury in CDH. According to this hypothesis, initial compromise occurs during the organogenesis stage resulting in bilateral hypoplasia, followed by ipsilateral lung compression secondary to herniation of the abdominal viscera at a later stage. This theory explains the variability of pulmonary hypoplasia on the contralateral side. The interference results in decreased branching of the bronchioles and pulmonary vessels causing acinar hypoplasia. Terminal bronchioles are reduced by thickening of the alveolar septa.8,9
The hypoplastic lung secondary to herniation of the viscera causes hypoplasia along with the pulmonary vessels. This results in reduced blood supply to the hypoplastic alveolar-capillary units. After the infant transitions from the fetal circulation, these effects are more pronounced resulting in pulmonary hypertension leading to right ventricular dysfunction. Secondary to pulmonary hypertension, there is a right-to-left shunting of blood across the patent foramen ovale and patent ductus arteriosus. Left ventricular dysfunction together with left atrial dysfunction leads to pulmonary venous hypertension and worsening of pulmonary arterial hypertension. It presents clinically in a wide spectrum from labile pre & postductal saturation to profound cyanosis.

To make a diagnosis of CDH, you can use an ultrasound examination during pregnancy. Prenatal diagnosis by ultrasound detects more than 50% of cases of CDH at a mean gestational age of 24 weeks. Performing prenatal diagnosis makes it possible to carry out patient counseling, referral to tertiary care centers, and sorting high-risk fetuses for fetal intervention. Direct signs of CDH in the fetus are the presence of abdominal organs in the thoracic cavity, and indirect signs include
polyhydramnios, shift of the cardiac axis, and shift of the mediastinum. Left-sided CDH is characterized by the presence of a fluid-filled stomach and small intestine in the thoracic cavity, close to the heart. Hepatic hernia appears as a homogeneous mass in the chest at heart level that continues with the intra-abdominal hepatic. Right-sided CDH is more difficult to diagnose because hepatic and pulmonary herniations have similar echogenicity. Identifying the gallbladder in the chest cavity is diagnostic of right-sided CDH. Approximately 40% of infants with CDH are not diagnosed prenatally and develop tachypnea, chest retraction, tachycardia, and cyanosis after birth. Physical examination may reveal a barrel-shaped chest, scaphoid abdomen, and absence of breath sounds on the ipsilateral side. Chest and abdominal radiographs are usually diagnostic and show a mediastinal shift with gas-filled stomach and bowel loops in the chest.

Intervention in CDH can be done prenatally or postnatally. The goal of fetal intervention in severe CDH is to prevent pulmonary hypoplasia and restore adequate lung growth to improve survival. Initial attempts at repair of the open fetal patch did not improve results and have since been abandoned. Furthermore, several animal studies have shown that prenatal tracheal occlusion (TO) prevents normal passage of lung fluids through the upper airways into the amniotic cavity, resulting in accelerated lung parenchymal growth with increased numbers of alveoli, capillaries, and lung remodeling. Pulmonary arterioles (Lung Pairs Until They Grow [PLUG]). However, prolonged TO reduces the number of type II pneumocytes, resulting in surfactant deficiency. Reversing TO before birth allows normalization of type II pneumocyte density (plug-unplug). Early human studies of prenatal TO were performed using temporary metal tracheal clips after maternal hysterotomy and fetal neck dissection. This was followed by an alternative approach involving maternal laparotomy and endoscopic fetal TO. However, subsequent randomized controlled trials (RCTs) showed no benefit, and this technique is no longer in practice. The only fetal intervention currently offered for CDH is fetal endoluminal TO (FETO), which involves percutaneous fetoscopic-assisted tracheal balloon placement at 27–29 weeks GA for severe cases of isolated CDH with O/E LHR <25% and liver herniation. Early human studies of prenatal TO were performed using temporary metal tracheal clips after maternal hysterotomy and fetal neck dissection. This was followed by an alternative approach involving maternal laparotomy and endoscopic fetal TO. However, subsequent randomized controlled trials (RCTs) showed no benefit, and this technique is no longer in practice.
which involves percutaneous fetoscopic-assisted tracheal balloon placement at 27–29 weeks GA for severe cases of isolated CDH with O/E LHR <25% and liver herniation. At birth, infants with CDH or suspected CDH should be placed on an orogastric/nasogastric tube with suction to achieve bowel decompression. Bag-mask ventilation should be avoided. Most of these infants (especially those with a prenatal diagnosis of CDH) require intubation in the delivery room. A pre-ductal pulse oximeter was placed in the right upper extremity as soon as possible. Oxygen saturation targets are based on NRP guidelines. Ventilation using a T-piece resuscitation is preferred to avoid high airway pressures. Parameters of the ventilator as shown in the figure. Predectal blood gases and invasive blood pressure monitoring are preferred. Inhaled nitric oxide is often used for the management of PPHN. For blood pressure management, fluid boluses and vasopressor agents are used based on the parameters in figure 3.8

CDH repair can be achieved through a thoracic or abdominal approach, and can be done in an open or minimally invasive way. Long-term results depend, perhaps most importantly, on the characteristics of the diaphragm defect. Patients with predictable small muscle defects typically have negligible rates of recurrence and complications. There is an increasing trend of thoracoscopic repair, which is thought to minimize postoperative pain and scarring and speed recovery. Some series have shown a higher rate of recurrence via the thoracoscopic approach, although this may be biased by a higher rate of anatomic unfavorability and an inherent learning curve. Patients at the other end of the spectrum, with diaphragmatic agenesis, uniformly requires patch placement to close the diaphragm defect. These are usually made from synthetic materials but recently there has been growing interest in combining synthetic materials with additional biological coatings in an effort to support repair and promote growth of native tissue for long term stability. Finally, several groups extol the benefits of autologous, flap-muscle closure of the defect.
The prognosis for isolated CDH is generally better than for CDH complicated by multiple anomalies. ERNICA provides several prenatal ultrasound assessment points that can be used as predictors of fetal outcome. Among others are:10

A. Lung size10

1. The lung contralateral to CDH is measured in the axial plane at the level of the 4-chamber view of the heart. To get the most accurate measurements, the lungs should ideally be close to the ultrasound transducer.
2. To avoid shadowing, the transducer must be level with the fetal ribs, and therefore, a maximum of 2 ribs must be visible.
3. The contralateral lung area is measured using a tracking method that allows accurate area estimation even if the lung shape is irregular. The anterior-posterior diameter method is the least reproducible and should therefore not be used.
4. This gives the area of the lung in square millimeters. Then divided by head circumference (in millimeters) to get the LHR index case
5. To get o/e LHR=(Observed LHR/Expected LHR) x 100. Survival is almost 100% with O/E LHR>45%, 50% with LHR 25–45% and poor with LHR<25%.11

Table 1. Predictors of antenatal ultrasound for survival in CDH.8

<table>
<thead>
<tr>
<th>A. LHR is calculated by dividing the fetal lung area (mm²) by the fetal head circumference (mm). The area of the fetal lung is usually measured at a four-chamber view level by multiplying the longest diameter of the contralateral lung by its longest perpendicular diameter. As an alternative, some obstetricians trace the lung margins and measure the lung area. The fetal head circumference is measured by its longest electronic ellipse.</th>
</tr>
</thead>
<tbody>
<tr>
<td>a. LHR &gt; 1.35 associated with 100% survival</td>
</tr>
<tr>
<td>b. LHR 1.35-0.6 associated with 61% survival</td>
</tr>
<tr>
<td>c. LHR &lt; 0.6 - no survival</td>
</tr>
<tr>
<td>B. The observed LHR to the expected (O/E LHR) is calculated by dividing the observed LHR by the expected ratio for gestational age</td>
</tr>
<tr>
<td>a. Fetal lung area increases 16-fold compared with a 4-fold increase in head circumference between 12 and 32 weeks of gestation</td>
</tr>
<tr>
<td>b. O/E LHR &lt; 25% is considered severe CDH (10% survival with elevated liver and 25% with decreased liver)</td>
</tr>
<tr>
<td>c. O/E LHR &lt; 15% with elevated heart – 100% mortality.</td>
</tr>
<tr>
<td>C. Liver position (or presence of liver herniation)</td>
</tr>
<tr>
<td>a. Liver herniation with LHR &lt; 1.0 – 60% mortality</td>
</tr>
<tr>
<td>b. Liver in thorax – 56% survival;</td>
</tr>
</tbody>
</table>

Figure 4. Section Ultrasound at 4-chamber view level for lung area measurement.
Liver herniation (liver-up) is associated with a worse prognosis. Previous studies have reported 100% survival without hepatic herniation (descended liver) compared to 56% with hepatic herniation. Survival decreased from 74 to 45% with hepatic herniation as reported by a meta-analysis. Sagittal scan through the fetal body allows visualization of diaphragmatic defects. On the sagittal side of the lesion like that could indicate that there is a portion of the liver that has herniated into the thorax. Additional indirect signs that can help determine the position of the liver include the following:

1. Doppler interrogation of the hepatic vessels in the longitudinal or coronal plane: visualization of these vessels flowing over the diaphragmatic rim is diagnostic of hepatic herniation.
2. Visualization of the ductus venosus and gallbladder: the presence of any of these structures into the thorax is diagnostic of a herniated liver. Liver herniation is also suspected when these organs are ectopic in the abdomen.
3. Visualization of the left umbilical segment of the portal vein on color Doppler imaging in the axial plane at the level where abdominal girth is usually measured: deviation (bend) of this vessel towards the left side indicates a herniated liver.
4. Gastric position also indirectly supports the diagnosis of liver herniation.
C. Abdominal position

Evaluation of abdominal position was recently introduced as an indirect method for estimating disease severity in left-sided CDH, as it has been shown to correlate with the proportion of intrathoracic hearts as determined by MRI. In several case series, precise positioning from stomach to chest has been shown to correlate with postnatal mortality and morbidity independent of o/e LHR.

1. Grade 1 or abdomen: the stomach is in a normal position so that it is not visible on the chest
2. 2nd degree or left anterior across the chest: the abdomen is visualized anteriorly at the apex of the heart, in contact with the anterior chest wall
3. Grade 3 or mid-to-posterior left chest: abdomen not in contact with left anterior chest wall; but next to the atrial-ventricular heart valves but with the larger part still anterior
4. 4th degree or retrocardiac: most of the stomach is located posterior to the atrial-ventricular valve, next to the left atrium of the heart in the right chest
Figure 7. Schematic images (A) and typical ultrasound images (B) of a fetus with left-sided CDH and variable degrees of abdominal position

V. CONCLUSION

Congenital diaphragmatic hernia (CDH) is a developmental disorder in the form of a defect in the diaphragm which allows the abdominal organs to herniate into the thoracic cavity through the diaphragm. To make a diagnosis of CDH, you can use an ultrasound examination during pregnancy. Prenatal diagnosis by ultrasound detects more than 50% of cases of CDH at a mean gestational age of 24 weeks. Performing prenatal diagnosis makes it possible to carry out patient counseling, referral to tertiary care centers, and sorting of high-risk fetuses for fetal intervention. Treatment for CDH can be carried out antenatally by installing prenatal tracheal occlusion (TO), but this procedure is no longer used and has been replaced with fetal endoluminal TO (FETO). At birth, CDH patients need to have a nasogastric tube inserted or even ventilated according to their clinical condition. The patient was then considered for surgical management to repair CDH. The prognosis of CDH patients can be determined using antenatal ultrasound with several indicators such as lung-to-ratio (LHR), hepatic herniation and abdominal position. Survival is nearly 100% with O/E LHR>45%, 50% with LHR 25–45% and poor with LHR<25%. Liver herniation (liver-up) is associated with a worse prognosis.

REFERENCES


