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Case Report

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Diagnosis of Congenital Diaphragmatic Hernia

A. Pegkou, Z. Fasoulakis, K. Tasias, M.I. Chatziioannou, G. Asimakopoulos, M. Theodora, G. Daskalakis, P. Antsaklis

Department of Obstetrics and Gynecology, Alexandra Hospital, National and Kapodistrian University of Athens - Faculty of Medicine, Athens, Greece

Corresponding Author

Zacharias Fasoulakis, 1st Department of Obstetrics and Gynecology – National and Kapodistrian University of Athens, Lourou 2-4, 115 28, Athens, Greece, email: hzaxos@gmail.com

Abstract

Background: Congenital Diaphragmatic Hernia (CDH) is a life-threatening condition occurring in newborns, characterized by a defect in the diaphragm that allows abdominal organs to migrate into the thoracic cavity. This report presents a case of CDH, outlining its diagnosis and highlighting the importance of early detection and management. We describe a case of a prenatal diagnosed left-side CDH that underwent karyotype screening with parents chosing termination of pregnancy. This case underscores the critical need for early diagnosis of CDH. In our review, we emphasize the role of prenatal ultrasound and MRI in early detection and evaluation of CDH, the importance of delivery in a specialized center capable of providing high-level neonatal care, and the need for ongoing research to improve prognosis and manage long-term complications associated with CDH.

Key words: Congenital diaphragmatic hernia, case report, prenatal diagnosis, ultrasound, MRI

Introduction

Congenital Diaphragmatic Hernia (CDH) is a defect that results from incomplete development and closure of the diaphragm at 1st trimester of pregnancy. This defect allows the contents of the abdominal cavity to migrate into the chest and causes compression of the lungs and the heart, leading to abnormal lung development, pulmonary hypoplasia, and underdevelopment of the ventricles of the heart[1-3]. CDH is a rare condition with an incidence of 1/2200, the majority of which (85%) are left-sided, but can also appear on the right side or rarely on both sides of the chest. Usually, it is prenatally diagnosed, especially in the second or third trimester, but some of them remain undiagnosed until birth[4-7].

Case presentation

A 22-year-old woman (G1P0) at 12 5/7 weeks of gestation presents for chorionic villus sampling due to findings on routine ultrasound of the first trimester that had been done at another diagnostic center. On ultrasound before the reception, the heart seemed to be more to the right than expected

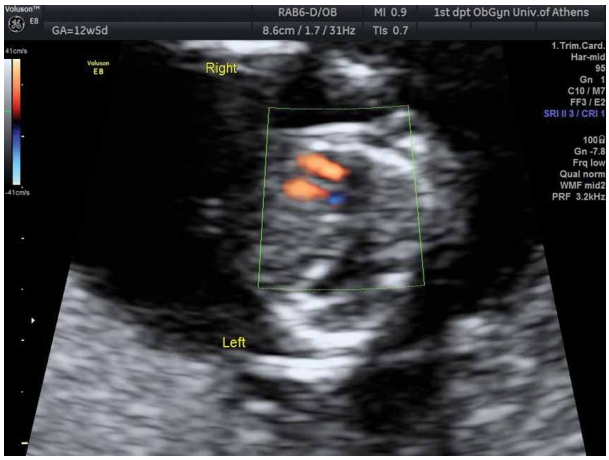

Figure 1. Heart shift to the right.

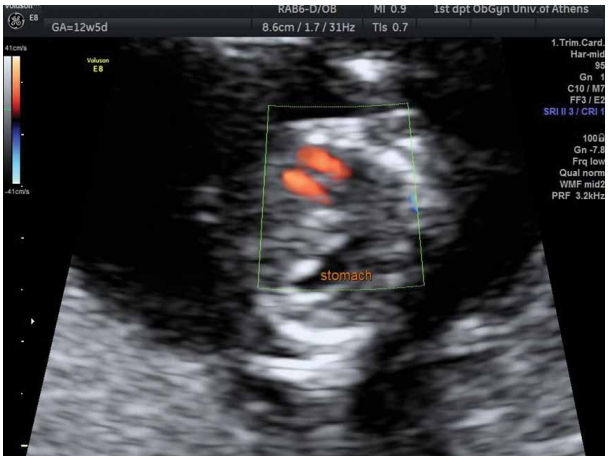

Figure 2. Heart shift to the right.

(Figure 1 & 2). The ductus venosus waveform was normal (Figure 3). A new ultrasound examination was recommended in a week. After a week, at 13 5/7 weeks of gestation, on the new ultrasound, the heart was displaced to the right and the fetal stomach was seen in the chest (Figure 4-6). The findings were diagnostic of left-sided congenital diaphragmatic hernia (CDH). The karyotype testing showed a normal genotype. The termination of pregnancy was chosen, after discussion and informing the parents about the poor prognosis and the high perinatal mortality rates.

Discussion

CDH is a severe anomaly characterized by a defect in the diaphragm, permitting the abdominal organs to herniate into the thoracic cavity, thus hindering the development of the lungs[3]. This life-threatening condition often results in significant morbidity and mortality, chiefly due to pulmonary hypoplasia and

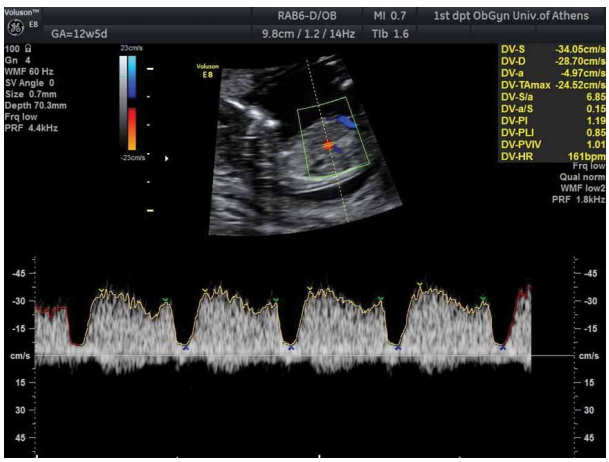

Figure 3. Ductus venosus.

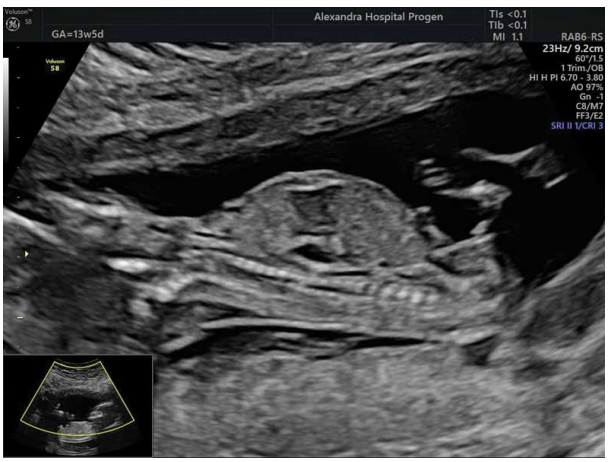

Figure 4. Fetal stomach into the chest.Fig.5 Heart shift to the right.

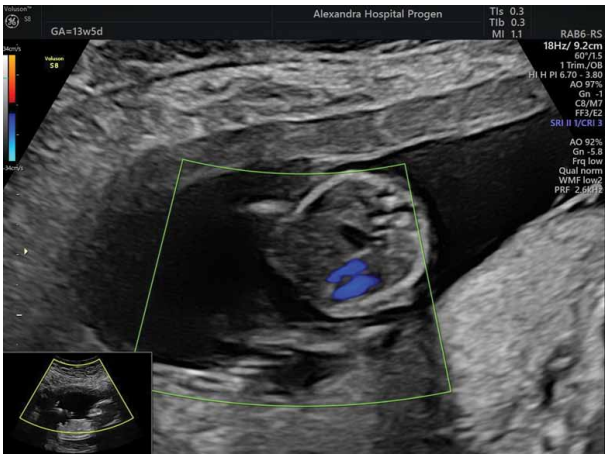

Figure 5. Heart shift to the right.

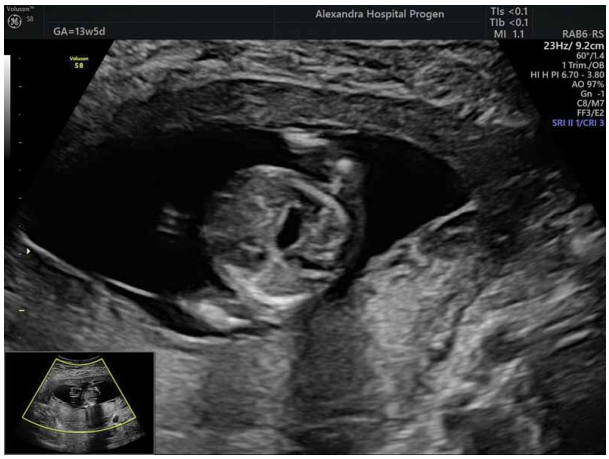

Figure 6. Stomach into the chest.

pulmonary hypertension[4].

The timely diagnosis of CDH is crucial as It guides the management approach. Prenatal diagnosis is generally achieved through ultrasound screening[10]. Ultrasonography allows the detection of herniated abdominal contents in the chest, and sometimes even of the defect itself. It also helps in understanding the severity of the condition by assessing parameters such as lung-to-head ratio (LHR) and position of the liver. Nonetheless, the severity of pulmonary hypoplasia and hypertension can only be truly ascertained Postnatally, necessitating careful monitoring and swift postnatal management[7].

A study by Jani et al. (2006) noted that an early prenatal diagnosis (before 25 weeks) significantly correlates with a poorer prognosis than a later diagnosis. This was attributed to the probability that an early diagnosis indicates a larger hernia and thus a more severe degree of pulmonary hypoplasia[2].

MRI is emerging as a complementary diagnostic tool in the assessment of CDH. According to a study by Victoria et al. (2014), MRI provides superior soft tissue contrast and a larger field of view, permitting a detailed assessment of lung volumes and associated anomalies[9].

Management options for CDH include surgical correction postnatally or fetoscopic tracheal occlusion (FETO) prenatally in severe cases. FETO aims to promote lung development by temporarily blocking the fetal trachea to create an environment of fluid buildup in the lungs. This stimulates lung growth, albeit with significant risks and complications[1].

Prompt diagnosis of CDH is pivotal for optimal perinatal management. While ultrasound remains the mainstay of prenatal diagnosis, other modalities like MRI can provide invaluable insights. Regardless of the management approach, multidisciplinary collaboration is key to optimizing outcomes for these complex patients.

In the last two decades, advancements in neonatal intensive care and the development of specialized centers for the management of CDH have improved the survival rates for this condition[8]. Despite this, long-term follow-up studies show a substantial proportion of CDH survivors suffer from long-term morbidity, including neurodevelopmental delay, hearing loss, and respiratory and nutritional problems[6].

Optimal management of CDH requires a comprehensive approach that starts with accurate prenatal diagnosis and evaluation, followed by delivery at a

tertiary center capable of providing extracorporeal membrane oxygenation (ECMO) support if necessary, timely surgical repair, and vigilant postoperative care. Moreover, lifelong follow-up is necessary to identify and manage the long-term complications of CDH[5].

In conclusion, CDH is a challenging condition that calls for continued research and clinical efforts to improve prenatal diagnosis, understand the genetic underpinnings, enhance surgical techniques, optimize postoperative care, and manage long-term complications.

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