First Trimester Cystic Hygroma: Herald to Early Diagnosis of Congenital Diaphragmatic Hernia

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The early diagnosis of in utero congenital diaphragmatic hernia (CDH) allows a thorough evaluation for other morbidities that may be associated with CDH. Our patient was referred to us with a fetus at 13 weeks gestational age with a thick nuchal translucency. Our team performed a transvaginal ultrasound that revealed a large cystic hygroma, a heart displaced to the right, and a heterogeneous mass with peristalsis in the left chest. The fetus was diagnosed with CDH. The patient received genetic counseling after which she requested and underwent chorionic villus sampling. The early diagnosis of CDH allows for a timely intervention with the improved management of pre-natal and post-natal care. Better neonatal management is pivotal in providing a multidisciplinary consultation approach in order to furnish accurate counseling and prognostic information for the patient. [*P R Health Sci J 2021;40:53-55*]

Key words: Congenital diaphragmatic hernia, Cystic hygroma, Early diagnosis

A congenital diaphragmatic hernia (CDH) is a defect found in the diaphragm that causes the contents of the abdominal cavity to move or be displaced into the thoracic cavity. The mass occupying effect of abdominal contents into the fetal chest prevents the fetal lungs from developing, leading to pulmonary hypoplasia. The defect can range from a small opening in to a complete absence of the diaphragm. Approximately 1,600 babies, or 1 in every 2,500 live births, are born with this defect each year. Ultrasound, a form of prenatal diagnosis, has helped detect over 50% of CDH pregnancies by the mean gestational age of 24 weeks (1). Once the defect has been diagnosed, 3-dimensional ultrasounds, fetal echocardiography, and fetal magnetic resonance imaging (MRI) can be used to assess the severity of the CDH. There have been reports of cases with increased nuchal translucency (NT) that were later diagnosed with a CDH (6,7).

Cardiac dysfunction and persistent pulmonary hypertension of the newborns are linked back to the pathophysiology of CDH. Cardiac, gastrointestinal, and genitourinary anomalies (in up to 50% of cases); Fryns syndrome (a multiple congenital anomaly), and chromosomal aneuploidies have been found to be associated with CDH in 16 to 37 % of cases (5). Trisomy 18, 13, and 21 are the most common chromosomal abnormalities associated with CDH. Monosomy X, tetrasomy12p, and tetraploidy 21 have also been associated with CDH (2). Environmental exposures, deficiencies in nutrition, and many genetic factors are among the possible etiologies of CDH. Although there have been advances in medical and surgical CDH treatments, infants with this defect often spend a long time in the hospital because their management requires a multi-disciplinary approach, and the morbidity rates remain high, ranging from 6 to 8% (2).

A left sided CDH may present with the finding of a complex mass in the left fetal chest that may contain the stomach and possibly the intestines and the spleen. There may be a shift or displacement of the fetal heart depending on the side where the defect is located, and the gastrointestinal content occupying the fetal chest. Isolated CDHs have a better prognosis than those diagnosed as part of a syndrome or that are present with multiple anomalies (3,4). There are many associated syndromes; the most common include pentalogy of Cantrell, Apert syndrome, Brachman–Cornelia De Lange syndrome and Beckwith–Wiedemann syndrome (2). Chromosomal abnormalities have been reported in 16 to 37% of cases. There is a 65% survival rate if the defect is isolated. Mortality rates vary from 7 to 57% if it is not isolated (5). The defects that appear later in pregnancy and those associated with "sliding" intraabdominal content also tend to have a better prognosis as there is less association with pulmonary changes.

Case report

We report the case of a 32-year-old primigravida who was referred to our practice at 13 1/7 weeks gestation after her obstetrician identified a thickened NT at the time of her

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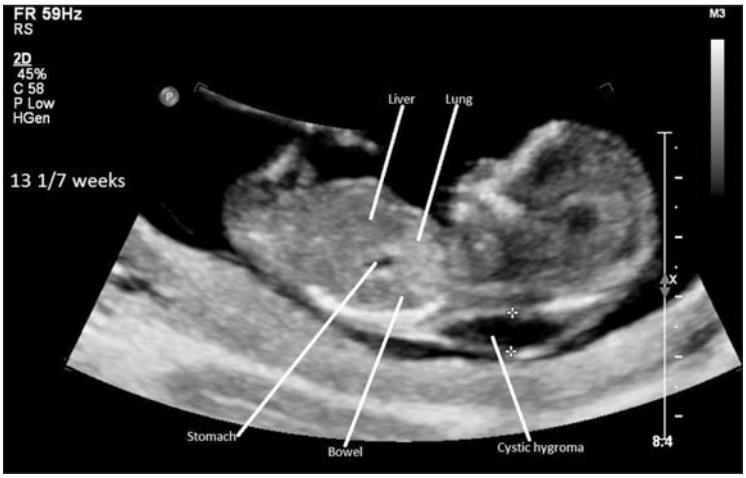

Figure 1. Sagittal view: Fetus at 13 1/7 weeks GA; showing increased nuchal translucency, with the intestines and stomach in the left fetal chest.

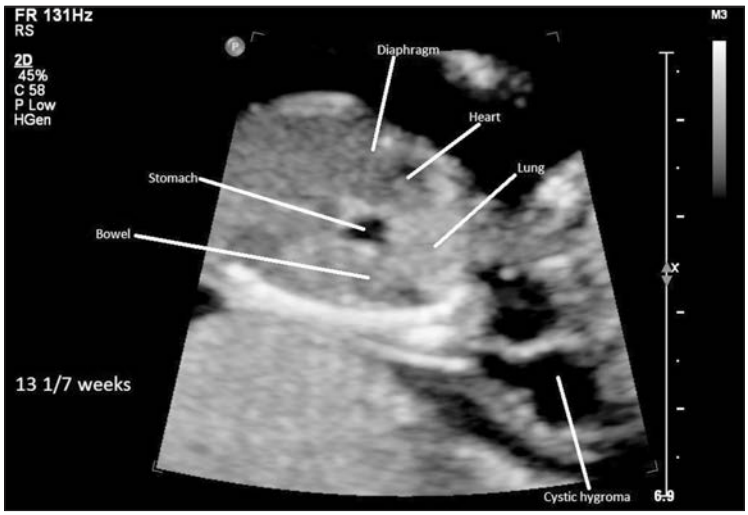

Figure 2. Sagittal view: Fetus at 13 1/7 weeks GA; showing a large cystic hygroma, with the intestines and stomach in the left fetal chest.

aneuploidy screening ultrasound. The patient was evaluated by our maternal–fetal medicine (MFM) team, which performed a transvaginal ultrasound and identified a large cystic hygroma with septations with an NT of 7.1 mm (>99th percentile), a heart displaced to the right and a heterogeneous mass with peristalsis on the left side of the chest. She underwent genetic counseling and chorionic villus sampling (CVS). CVS revealed a normal karyotype, microarray analysis, and a negative result for Di George and Noonan syndromes. The anatomy ultrasound at 16 3/7 weeks confirmed a left-sided CDH. The patient was managed with a multidisciplinary approach including an OB/GYN, an MFM team, neonatologists, geneticists, pediatric surgeons, and a social worker. An MRI done at 27 weeks confirmed a left-sided CDH containing bowel, the stomach and part of the liver with a resulting right mediastinal shift.

The frequency of the follow-up ultrasound evaluations was once a month until she reached 32 weeks in her pregnancy. She continued to have regular visits with her OB/GYN. The remainder of the pregnancy was uneventful until 34 weeks gestation, at which point the patient started experiencing premature contractions. She was hospitalized for evaluation. She went on to deliver at term. After the delivery and intubation, the newborn was admitted to the neonatal intensive care unit. The neonate required mechanical ventilation but no ECMO. He underwent a postnatal surgical repair of the CDH on day of life 15. The post-repair neonatal period was uneventful, with the biggest challenge being nutritional support. The neonate was discharged home on day of life 66. Nutritional challenges remained for the first year of the infant's life.

Discussion

Diagnosis in utero, allows for a thorough evaluation to assess for other morbidities that may be associated with CDH as well as for early intervention with the improved management of pre-natal and post-natal care, which can lead to better neonatal management, family support and outcomes. Early diagnosis is pivotal in providing a multidisciplinary consultation approach including MFM specialists, fetal surgeons, geneticists, neonatologists and nutrition consultants in order to supply accurate counseling and prognostic information for these families. Once fully evaluated, these fetuses should be delivered at a tertiary care facility that can provide immediate respiratory support, as well as surgical intervention (as needed) and the use of ECMO.

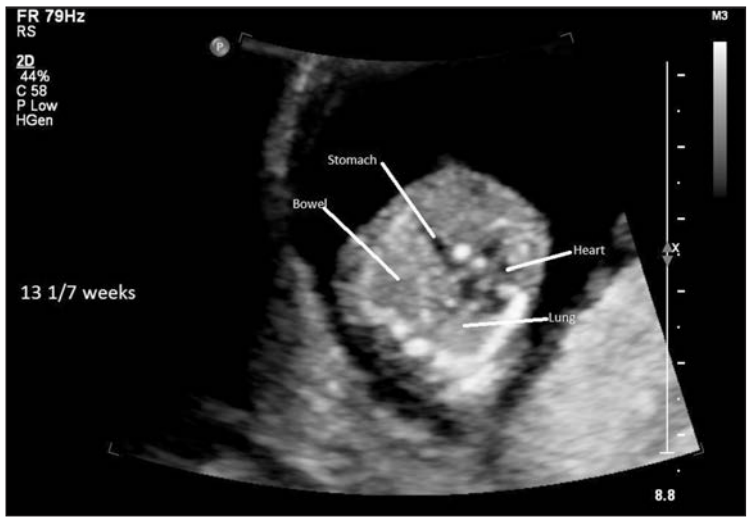

Figure 3. Coronal view: Fetus at 13 1/7 weeks GA; the heart is displaced to the right chest cavity, and the intestines and stomach are in the left fetal chest.

Resumen

El diagnóstico temprano en útero de la hernia diafragmática congénita (HDC) permite una evaluación completa de la morbilidad que puede estar asociada con la misma. La paciente se presentó a nuestra oficina con un feto a las 13 semanas de gestación con una translucencia nucal engrosada. Nuestro equipo realizó una ecografía transvaginal la cual reveló un higroma quístico grande, el corazón desplazado hacia la derecha y una masa heterogénea con peristalsis en el tórax izquierdo. El feto fue diagnosticado con HDC. En vista de los hallazgos, la paciente recibió consejería genética, luego de lo cual decidió someterse a un muestreo de vellosidades coriónicas. El diagnóstico precoz permite una intervención temprana con un mejor manejo de la atención prenatal y postnatal. Un mejor manejo neonatal es fundamental para proporcionar un enfoque de consulta multidisciplinaria con el fin de ofrecer un asesoramiento e información del pronóstico más precisos para la paciente.

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