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CONGENITAL DIAPHRAGMATIC HERNIA: PRENATAL DIAGNOSIS

Cristina-Crenguta Albu[1*], Adriana Vasilache[1], Mihaela Tanase[1], Ioana Andreea Stanciu[1], Dinu-Florin Albu[2] and Stefan-Dimitrie Albu[1]

[1]University of Medicine and Pharmacy "Carol Davila", 37 Dionisie Lupu Street, 1st District, 020021, Bucharest, Romania.

[2]Clinical Hospital of Obstetrics and Gynecology "Prof. Dr. Panait Sârbu", 5 Giulesti Street, 6st District, 060251, Bucharest, Romania.

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***Corresponding Author**
Cristina-Crenguta Albu
University of Medicine and Pharmacy "Carol Davila", 37 Dionisie Lupu Street, 1st District, 020021, Bucharest, Romania.

ABSTRACT

The congenital diaphragmatic hernia (CDH) is a most severe birth defect of the lung, associated with high neonatal mortality and a significant rate of morbidity. We are presenting here a isolated, non-hereditary giant congenital diaphragmatic hernia, associated with intrathoracic herniation of the stomach, the ileal loops, and the heart, severe pulmonary hypoplasia, and predicted survival of about 15%, successfully diagnosed prenatally by ultrasound examination at 24 weeks of pregnancy. Prenatal ultrasound scan was decisive in the early prenatal diagnosis and optimized management of the malformed fetus.

KEYWORDS: congenital diaphragmatic hernia, pulmonary hypoplasia, ultrasound examination, prenatal diagnosis.

INTRODUCTION

The congenital diaphragmatic hernia (CDH) is a most severe birth defect of the lung, associated with high neonatal mortality and a significant rate of morbidity due to associated pulmonary hypoplasia, pulmonary hypertension, and heart failure.[1-3]

The rate of CDH incidence is about 0.5 ‰ live births.[4] About 50% of cases are isolated CDH while 50% are syndromic or complex CDH, reflecting the interaction between genetic and non-genetic factors.

The complex CDH is usually associated with various types of chromosomal abnormalities (aneuploidies 13, 18, 21), autosomal recessive syndromes (Donnai–Barrow syndrome,

Matthew–Wood syndrome, BNAR syndrome, Cutis laxa, Spondylocostal dysostosis), autosomal dominant syndromes (Denys Drash syndrome, Meacham syndrome, Frasier syndrome, Marfan syndrome, SHORT syndrome, Apert syndrome), X linked syndromes (Focal dermal hypoplasia/Goltz– Gorlin, Craniofrontonasal syndrome, Lowe syndrome, Simpson–Golabi–Behmel syndrome) and congenital anomalies (cardiovascular and gastrointestinal abnormalities, neural tube defects, limb, genitourinary, and eye abnormalities).[5] The present study aims to illustrate the supremacy of ultrasound examination prenatal and the importance of early prenatal diagnosis in the prevention of severe congenital defects.

MATERIALS AND METHODS

A 34-year-old, primiparous caucasian woman, was referred in 24 weeks of pregnancy, in a private clinic from Bucharest, Romania, in February 2020, for a routine prenatal ultrasound examination of the fetal malformations.

The couple was non-consanguineous and clinically healthy, without hereditary or familial pathologies.

The ultrasound examination was performed with a General Electric Voluson E10 Ultrasound Machine.

After the pregnant woman was informed about the ultrasound investigation, with her informed consent for the sonogram, the scan was performed transabdominally by an experienced sonographer.

RESULTS

The ultrasound scanning highlighted a single fetus 24.3 weeks old (chronological) and 25 weeks old (biometric), in evolution, with an estimated fetal weight of 672 g.

A detailed second-trimester ultrasound examination of the fetus revealed several major abnormalities.

At the level of the cephalic extremity: the biparietal diameter was 62.7 mm; the occipitofrontal diameter was 79.7 mm and head circumference was 224.7 mm (Fig. 1 and Fig. 2).

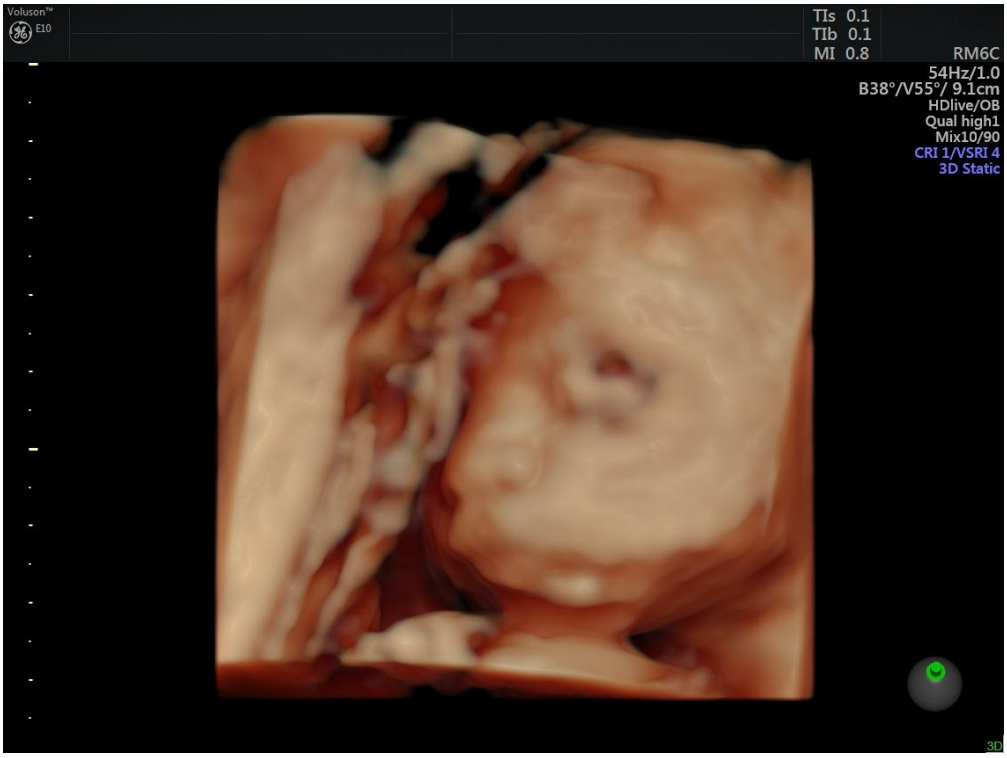

Figure 1: 3D Static ultrasound examination of the fetal face.

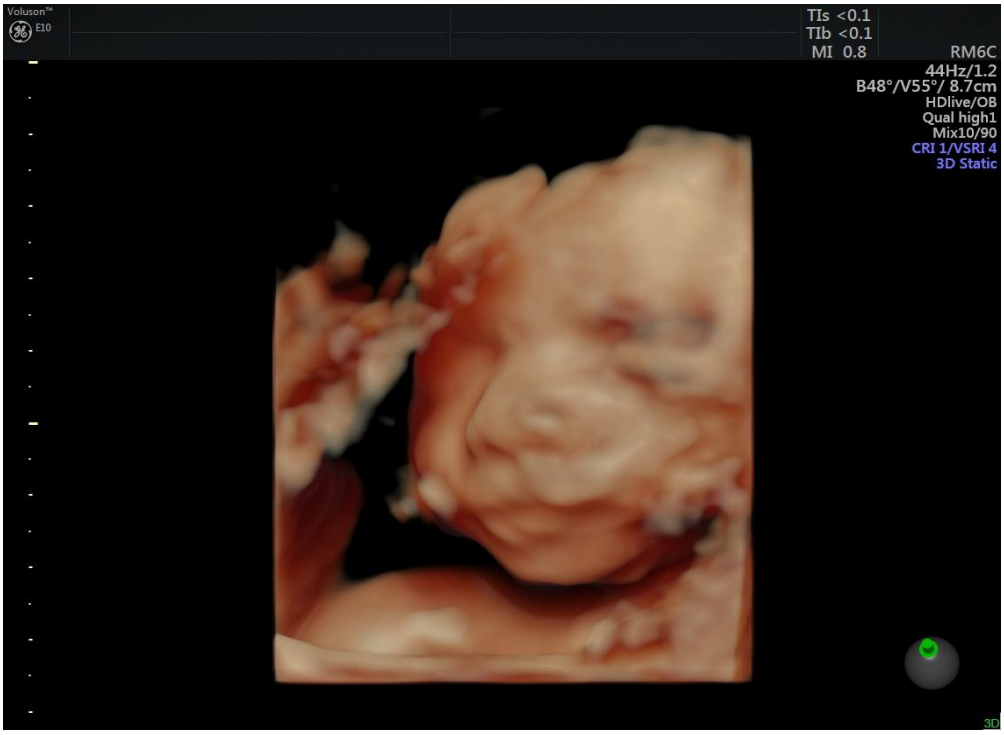

Figure 2 : 3D Static ultrasound imaging of fetal face with clear features.

At the level of the thorax: the anterior-posterior diameter was 66.4 mm and the transverse diameter was 52.5 mm, with significant structural abnormalities of the chest cavity. In the left hemithorax, a large congenital diaphragmatic hernia, with a size of 19.9 mm is revealed (Fig.

3). The fetal stomach is completely highlighted in the left hemithorax, together with the ileal loops, as a result of their penetration through the diaphragmatic defect (Fig. 4). Also, ultrasound examination highlights severe pulmonary hypoplasia, and fetal echocardiography pointed tetracameral heart located in the right hemithorax (Fig. 5). No other fetal dysmorphisms over 0.5 cm were found. In conformity with the fetal morphology examination, the following diagnosis was established: Mono-fetal pregnancy 24.3 weeks old (chronological) and 25 weeks old (biometric), in evolution. Estimated fetal weight 672 g. Malformed fetus: Giant congenital diaphragmatic hernia. Intrathoracic herniation of the stomach, the ileal loops, and the heart. Severe pulmonary hypoplasia.

The parents were informed about the major gravity of anomalies, the severity of pulmonary hypoplasia, with predicted survival of about 15%, and decide to terminate the pregnancy for medical reasons. Anatomopathological examination confirmed the prenatal diagnosis.

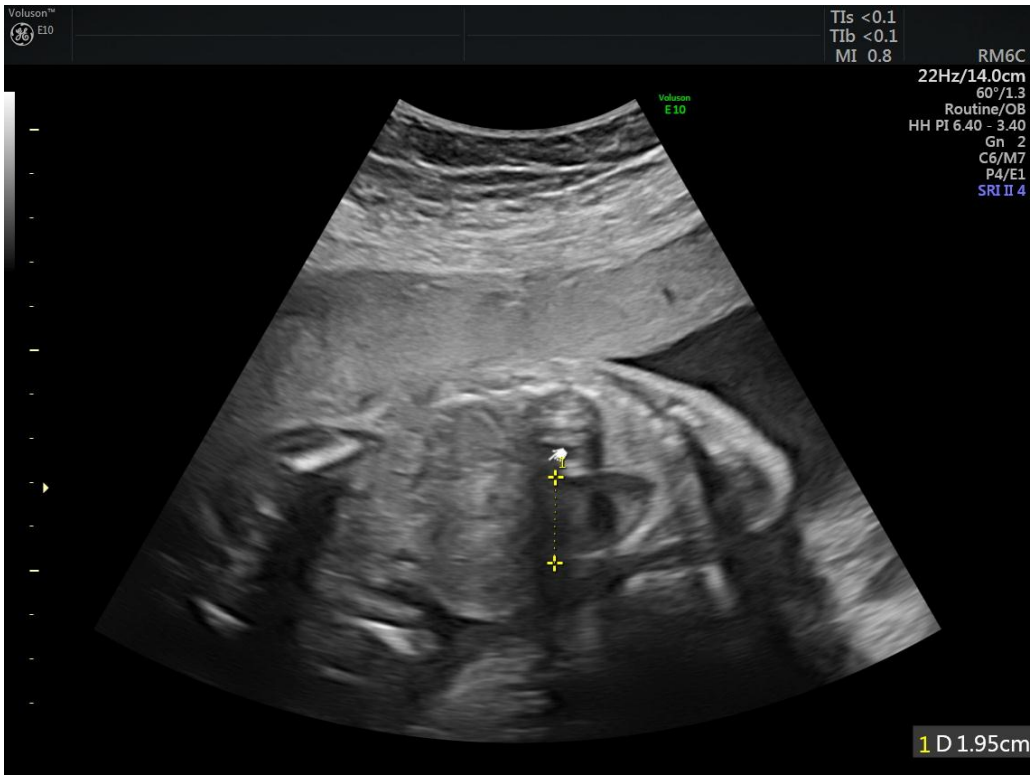

Figure 3 : 2D Fetal ultrasound examination.

Large congenital diaphragmatic hernia (1.95 cm) in the left hemithorax.

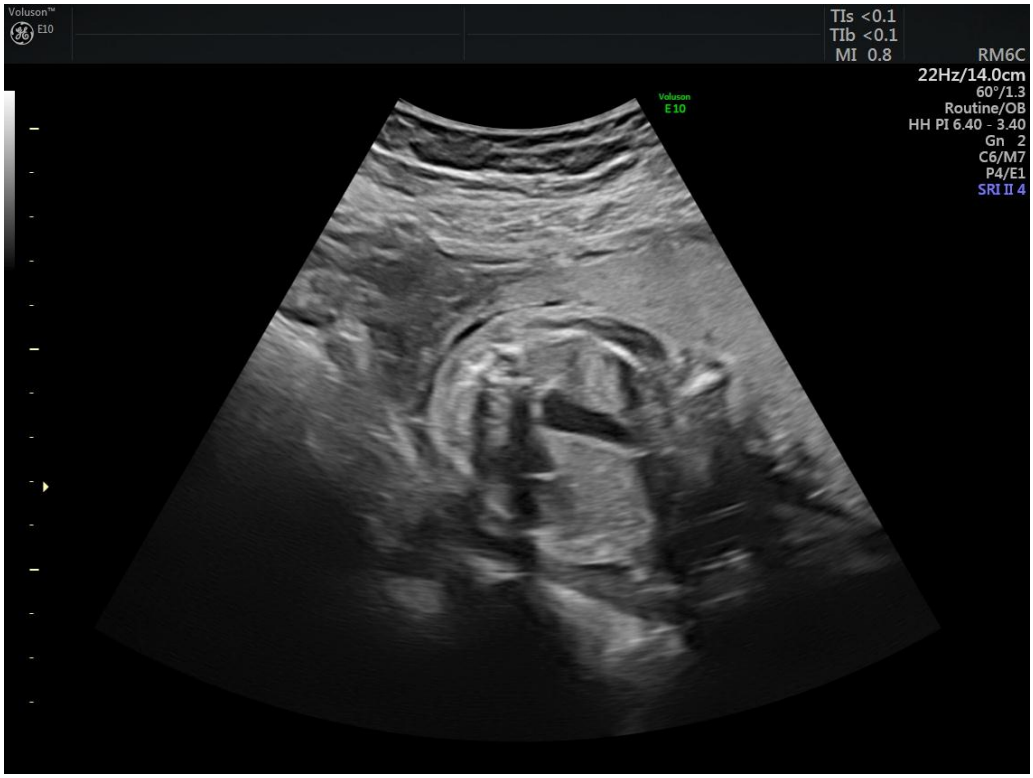

Figure 4: 2D fetal ultrasound examination: Congenital defect of the left hemidiaphragm with large diaphragmatic hernia.

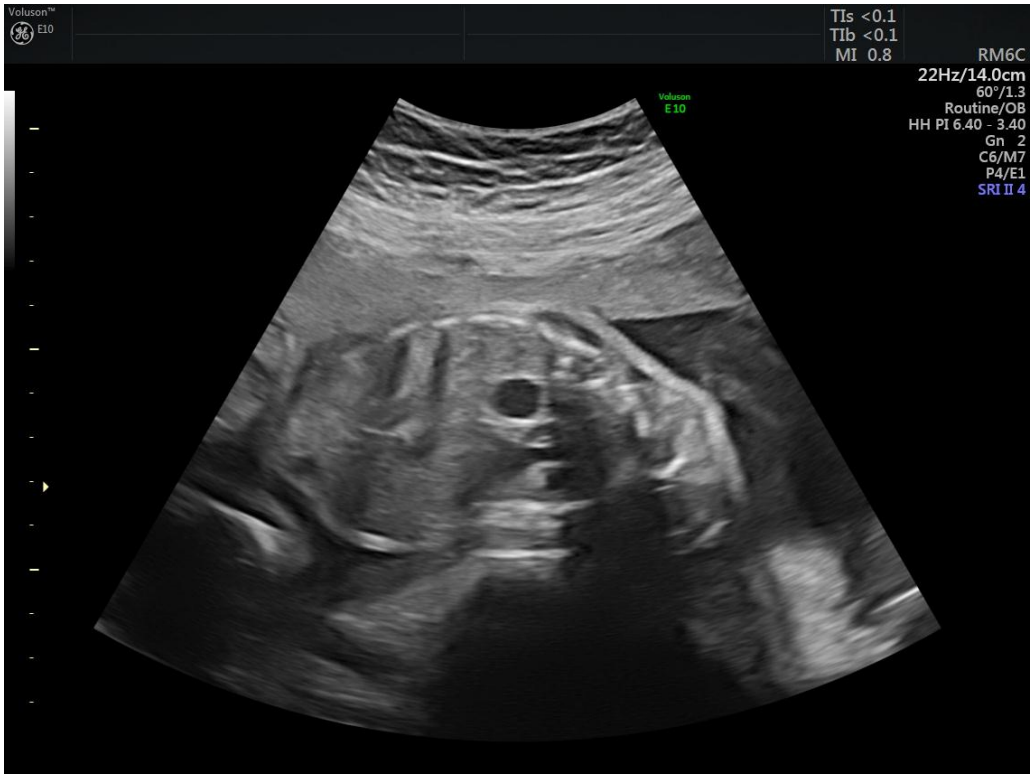

Figure 5: 2D Fetal ultrasound examination: Left-sided congenital diaphragmatic hernia.

DISCUSSION

CDH is a complex and unforgiving syndrome, one of the most dreaded congenital anomalies in the neonatal intensive care units because of a high mortality rate dependent upon multiple factors: the size of the hernia, organs involved, additional congenital malformation, chromosomal and genetic abnormalities, and complications.[6-10]

This condition can often be diagnosed before birth by prenatal ultrasonography at a mean gestational age of 24.2 weeks.

Genetic counseling after prenatal diagnosis of complex structural birth defects is very difficult because of the bad prognosis, but the early prenatal diagnosis of extremely severe birth defects is crucial for the prevention of neonatal mortality and morbidity.[11-14]

In our research, the prenatal ultrasound diagnosis of was crucial, because it was possible to avoid the appearance of a newborn severely affected congenitally, through an accurate prenatal diagnosis.[14]

CONCLUSION

In our case, prenatal ultrasound examination was decisive in the early prenatal diagnosis and optimized management of the malformed fetus with a giant congenital diaphragmatic hernia with intrathoracic herniation of the stomach, the ileal loops, and heart, associated with severe pulmonary hypoplasia, and predicted survival of about 15%.

Authors' contributions

All authors contributed equally with the first-author, in the preparing, review and editing of the article. All authors read and approved the final version of the manuscript.

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