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Case report

Prenatal diagnosis of right-sided congenital diaphragmatic hernia

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Abstract

This case report details the prenatal diagnosis and management of a right-sided congenital diaphragmatic hernia (CDH) in a 37-year-old pregnant woman. In the 22nd week of the pregnancy, a screening ultrasound revealed an abnormal mass in the right fetal chest, suspected to be a diaphragmatic hernia. A series of fetal ultrasound and echocardiographic examinations were conducted, revealing a mass of intestines in the right chest displacing the heart (with normal heart anatomy). Fetal magnetic resonance imaging at 27 weeks confirmed the diaphragmatic hernia. Subsequent monitoring revealed unfavourable prognostic indicators, including a decreased lung-to-head ratio (LHR) index. Premature rupture of membranes occurred in the 28th week. An urgent caesarean section was performed at 31 weeks due to the onset of labour and signs of fetal distress, and a female newborn with a birth weight of 1800 g was delivered. Right-sided diaphragmatic hernia was confirmed postnatally. Surgical intervention was undertaken to relocate the intestines and close the diaphragmatic defect. Despite initial efforts, the newborn's respiratory and circulatory function deteriorated, leading to the unfortunate demise on the sixth day of life.

Key words: magnetic resonance imaging, prenatal diagnosis, fetal echocardiography, fetal ultrasound, right-sided congenital diaphragmatic hernia.

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Introduction

Congenital diaphragmatic hernia denotes an anomalous development of the fetal diaphragm, resulting in the displacement of the intestines, liver, and/or stomach into the thoracic cavity. This condition is concomitant with fetal lung hypoplasia and pulmonary hypertension. We present a case report of prenatally diagnosed right-sided congenital diaphragmatic hernia (RSCDH), which is a much less common variant of this defect and our diagnostics dilemma comparing ultrasound versus magnetic resonance imaging (MRI) data.

Case report

The patient was a 37-year-old pregnant woman. This was her fourth pregnancy; the first ended with a vaginal delivery of a full-term newborn, the next 2 ended with spontaneous miscarriages in the first trimesters. The last miscarriage

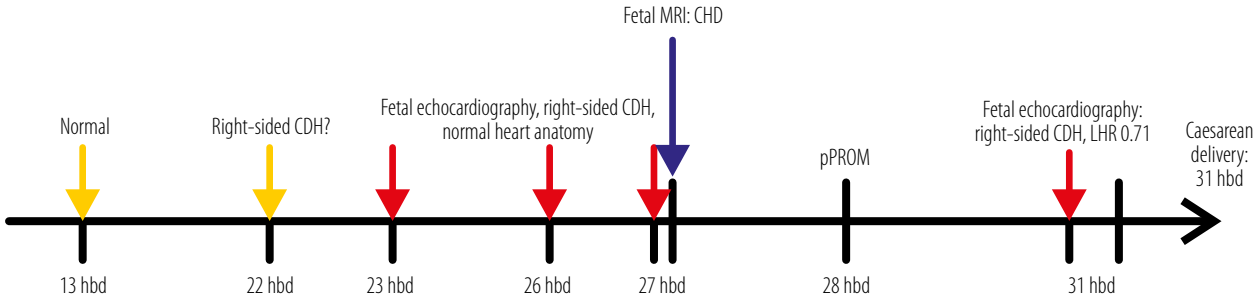

Figure 1. Diagram of the fetal diagnostic process

Yellow arrows – screening ultrasound, red arrows – fetal echocardiography, purple arrow – fetal MRI.

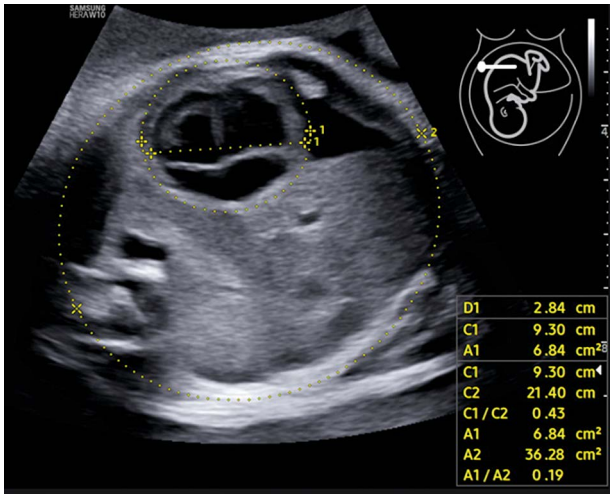

Figure 2. Fetal heart size below normal. The HA/CA (heart area/chest area) index value is below 0.2

occurred 3 months before the current pregnancy. The pregnant woman was obese (BMI 33). In the first trimester of pregnancy, she was diagnosed with gestational diabetes, which was initially treated only by diet and since the 26th week of pregnancy required insulin therapy.

No fetal abnormalities were detected in the screening obstetric ultrasound examination in the 13th week of pregnancy. In an anomaly-scan ultrasound examination in the 22nd week of pregnancy, in this high-risk pregnancy, the obstetrician found an abnormal mass in the right part of the fetal chest and a shift of the heart to the left side of the chest. Suspected of diaphragmatic hernia, he referred the pregnant woman to our referral centre.

A total of 4 fetal ultrasound + echocardiographic examinations were performed in our centre (Figure 1). Based on ultrasound evaluation the fetus was found to have a pathological mass with echogenicity resembling intestines in the right part of the chest. This mass moved the heart to the left side and compressed it. The value of the heart area/chest area (HA/CA) ratio was 0.19 (below normal) (Figure 2). The fetal stomach was visible or in fetal chest or below the diaphragm (Figure 3). There were no structural or functional changes in the fetal heart. Circulatory system efficiency was assessed at 8/10 points on the CVPS scale (–2 points for heart size below normal). A normal fetal growth trend was observed in all studies. The volume of amniotic fluid remained within normal limits

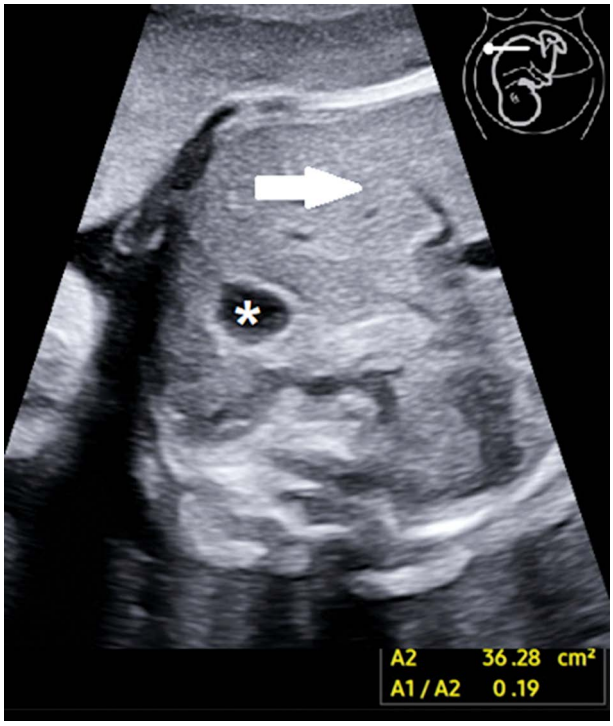

Figure 3. The liver (white arrow) and stomach (white asterisk) of the fetus are visible below the diaphragm

(AFI 17-22 cm). Flow spectra in the umbilical arteries and middle cerebral arteries were normal. Fetal magnetic resonance imaging was performed in the 27th week of pregnancy. The magnetic resonance imaging (MRI) confirmed the presence of diaphragmatic hernia in the fetus with displacement of the intestines into the chest (Figure 4). At 26 weeks of pregnancy, prenatal steroid therapy (betamethasone) was administered, and a consultation was scheduled to consider fetal balloon tracheal occlusion (FETO). However, before the consultation took place, the premature rupture of membranes occurred in the 28th week of pregnancy. In the Department of Obstetrics and Gynaecology, broad-spectrum antibiotic therapy was implemented, laboratory markers of inflammation were controlled, and the well-being of the fetus and the pregnant woman was monitored. The last fetal ultrasound and fetal echocardiographic examination was performed in the 31st week of pregnancy. The absolute value of the LHR index was 0.71, which was 36% of the expected value for this week of pregnancy. These values suggested an unfavourable prognosis. Ad-

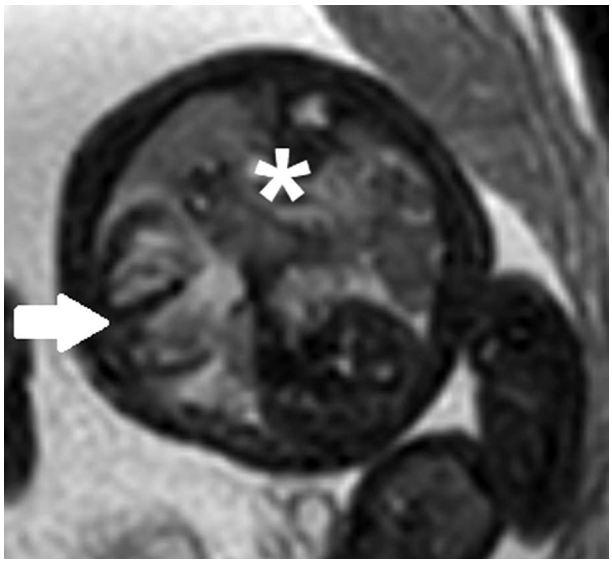

Figure 4. Fetal MRI showing displacement of the intestines (white asterisk) to the right part of the chest and a shift of the heart (white arrow) to the left (radiologist: Dobromiła Barańska)

Table 1. Comparison for gender, coexisting anomaly, Apgar score, and mortality between left-sided CDH and right-sided CDH according to Abramov *et al.* [7]

Parameter	Left CDH	Right CDH
Number of cases	3156 (84%)	598 (16%)
Gender	60% males	60% males
Proportion of larger defects	31%	43%
Incidence of non-repair hernia	16%	21%
Coexisting one congenital anomaly	26.5%	28%
APGAR at minute 1	5.20	4.77
APGAR at minute 5	7.3	6.6
Mortality	28%	33%

CHD – congenital diaphragmatic hernia.

ditionally, slight protosystolic tricuspid valve regurgitation and small pericardial effusion (< 2.5 mm) were found. Circulatory system efficiency was assessed at 7/10 on the CVPS scale. Estimated fetal weight and peripheral Doppler flows were normal.

Five days after the last ultrasound and echocardiographic examination, the pregnant woman experienced regular uterine contractions leading to shortening and dilatation of the cervix. Due to fetal tachycardia of 200/min detected in the CTG examination, the impending intrauterine fetal asphyxia was suspected, and delivery was performed by emergency caesarean section.

The birth weight of the female newborn was 1800 g. The Apgar score was 5/6/6/7, and the blood pH was 7.084. On the first day after birth, the newborn's condition was serious, mechanical ventilation was implemented with pressor amines. An X-ray examination in the newborn confirmed a RSCDH with displacement of the intestines into the chest (Figure 5). The newborn was diagnosed with anaemia (haemoglobin 11 g%, haematocrit 33%), and RBC was transfused.

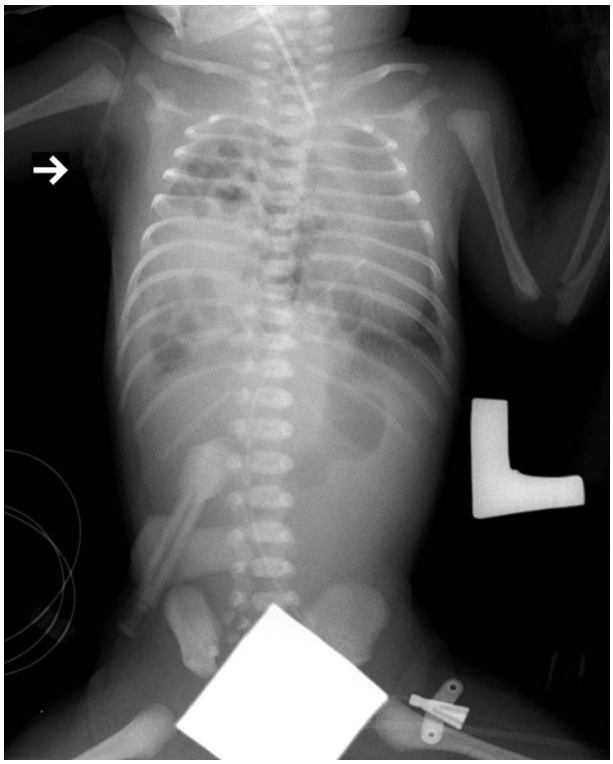

Figure 5. Chest X-ray of a newborn on the first day of life: displacement of the intestines to the right part of the chest and a shift of the heart to the left

Escherichia coli bacteria were cultured from the blood culture. The ECHO examination confirmed the normal structure of the heart and showed signs of pulmonary hypertension. On the second day of life, a surgical procedure was performed to divert the intestines from the chest into the abdominal cavity and close the defect in the diaphragm. On the sixth day of life, the deterioration of respiratory and circulatory efficiency occurred, with no response to treatment attempts. The newborn was pronounced dead on the same day. At the parents' request, an autopsy was not performed.

Discussion

The presented case involved a high-risk pregnancy that commenced 3 months after a prior miscarriage. Research by Strzelecka *et al.* indicates that, in instances of poor outcome of previous pregnancy (miscarriage or congenital heart defects in the fetus), maintaining an interval of at least 24 months between pregnancies may increase the probability of normal outcome in a subsequent pregnancy [1].

Evidence reveals that cases of congenital diaphragmatic hernia (CDH) with concurrent defects are more prevalent than isolated CDH cases. Between 1980 and 2009 in Europe, CDH with concurrent defects occurred at a rate of 2.3 per 10,000 births, while isolated CDH occurred at a rate of 1.6 per 10,000 births [2]. The primary concurrent defects include heart abnormalities, which align with a similar timeline of diaphragm development. Additional defects encompass craniofacial, central nervous system, and chromosomal abnormalities [2-4]. The co-occurrence of defects heightens the risk of mortality,

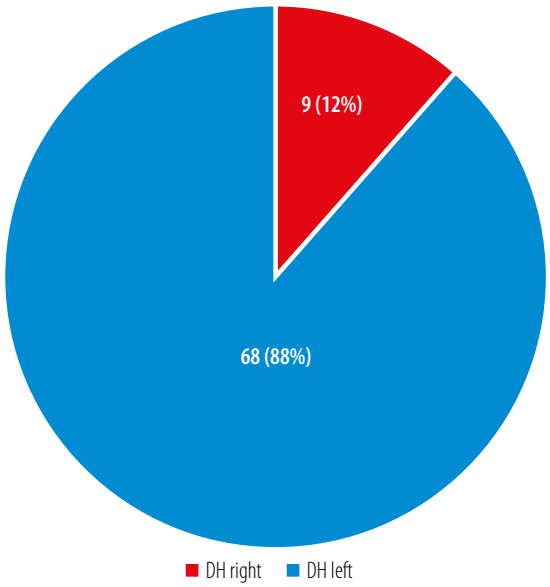

Figure 6. Proportion of left-sided congenital diaphragmatic hernia (CDH) and right-sided CDH cases between 2018 and 2023 in the Polish Mother's Memorial Hospital in Lodz

with a 100% mortality rate associated with central nervous system and heart defects [3].

Left-sided diaphragmatic hernia prevails in fetal cases, with right-sided occurrences being less frequent. RSCDH are linked to a higher mortality rate, often attributed to larger defect sizes. Research also indicates that the defect's size, rather than its lateralisation, plays a more pivotal role in determining the prognosis [5-7] (Table 1).

Between 2018 and 2023, at the Polish Mother's Memorial Hospital in Lodz (Poland), 9 newborns were born with congenital right-sided diaphragmatic hernia. This represented 12% of all newborns born with CDH in these years (Figure 6). In 77.8% (7/9) of cases, it was an isolated defect. In 22.2% (2/9) of cases, there was an accompanying congenital heart defect. The mean gestational age was 34.3 weeks (SD ±3.5). Caesarean section delivery occurred in 88.9% (8/9) of cases. Female newborns accounted for 55.6% (5/9) and male newborns for 44.4% (4/9). The mean birth weight was 2109 g (SD ±523 g). The mean Apgar score at one minute of life was 4.4 (SD ±2.7). The mean Apgar score at 5 minutes of life was 5.9 (SD ±2.1). Neonatal mortality upon discharge from the hospital was 66.7% (6/9). Newborns who survived until hospital discharge were born on average at 36.7 (SD ±2.1) weeks of gestation with a mean birth weight of 2317 g (SD ±293 g). Newborns who died before hospital discharge were born on average at 33.2 (SD ±3.6) weeks of gestation with a mean birth weight of 2005 g (SD ±604 g) (Table 2).

The congenital diaphragmatic hernia diagnosis relies on ultrasound scans, with the average gestational age at diagnosis being 22-24 weeks. Magnetic resonance imaging with lung volume assessment may enhance diagnostic accuracy, overcoming limitations posed by maternal obesity or oligohydramnios and providing superior soft tissue contrast [8]. Right-sided hernias pose a diagnostic challenge due to ultrasound imaging characteristics [5, 9-11] (Table 3).

Accurate diagnosis of congenital diaphragmatic hernia and determination of the defect's side necessitate proper identification of the left and right sides of the fetal body. This involves discerning fetal position (longitudinal/oblique/transverse, cephalic/pelvic) and locating the fetal spine. For example, if the fetus is in a longitudinal cephalic position and its spine is directed towards the left side of the uterine cavity, the left side of the fetal body will be directed towards the bottom of the ultrasound screen. If the fetus is in a longitudinal breech position and its spine is directed towards the left side of the uterine cavity, then the left side of the fetal body will be directed towards the top of the ultrasound screen. Placement of a pictogram displaying the fetal position on the screen during ultrasound and echocardiographic examinations aids image analysis and helps reduce diagnostic errors [12].

Table 2. Characteristics of fetuses and newborns with RSCDH born in the Polish Mother's Memorial Hospital in Lodz between 2018 and 2023

No.	Year	Isolated CDH	CDH with cardiac malformation	CDH with extracardiac malformation	Gestational age at delivery	Caesarean section/ Vaginal delivery	Gender	Birth-weight	Apgar score (minute 1, 3, 5, 10)	Neonatal death
1	2018	+	–	–	34 weeks	CS	B	1850	4/3/5/5	+
2	2020	+	–	–	39 weeks	CS	B	2200	10/10	–
3	2020	+	–	–	35 weeks	CS	G	2400	5/6/6/6	+
4	2020	+	–	–	35 weeks	CS	B	2100	3/4/6/7	–
5	2021	+	–	–	36 weeks	Vaginal	B	2650	5/5/5/5	–
6	2022	+	–	–	35 weeks	CS	G	2100	3/4/6/6	+
7	2023	–	+	+	27 weeks	CS	G	1050	0/0/0/2	+
8	2023	+	–	–	31 weeks	CS	G	1800	5/6/6/7	+
9	2023	–	+	+	37 weeks	CS	G	2830	5/6/7/7	+

CHD – congenital diaphragmatic hernia, CS – caesarean section, G – girl, B – boy.

Table 3. Characteristic ultrasound features of diagnostic imaging in the prenatal period according to the literature

Left CDH	Right CDH
Liver herniation into the thorax 47.9% [5]	Liver herniation into the thorax 96.1% [5]
Stomach and/or gallbladder in contact with the bladder wall [11]	Abnormal position of the stomach: horizontal orientation of the stomach [10]
Herniation of the stomach and intestines are likely [10]	Herniation of the stomach and intestines are not likely [10]
Reductions in LV width, area, and mass, reduced aortic valve diameter [9]	Less severe LV hypoplasia than in left-sided CDH [9]
Polyhydramnios [10]	Polyhydramnios [10]
Abnormal position of the heart [11]	Abnormal position of the heart [11]

CHD – congenital diaphragmatic hernia.

Recent studies have identified specific genetic mutations associated with RSCDH, such as mutations in the GATA4 transcription factor. Genetic testing can help identify people at risk of developing RSCDH and facilitate early intervention and genetic counselling [13-15].

Hypoplasia of the developing heart in CDH is a well-documented observation, initially identified in post-mortem studies and subsequently confirmed by echocardiography analyses. In RSCDH, limited available data suggest a reduction in both fetal right ventricular and pulmonary arterial dimensions, with less severe left ventricular hypoplasia than in left-sided CDH [9]. Potential indicators of poor prognosis in CDH include a small diameter of the ascending aorta (AAo) and a high ratio of pulmonary artery diameter to ascending aorta diameter (MPA/AAo) [16].

The risk of preterm delivery is elevated when CDH is diagnosed in the fetus, estimated to be between 22 and 35%. Cohort studies posit that preterm birth amplifies the mortality rate in CDH-affected fetuses, with a higher risk for RSCDH [17, 18]. One potential risk factor for premature delivery is the presence of polyhydramnios due to oesophageal compression and fetal swallowing impairment [8].

The risk factors of poor outcome in CDH are summarised in Table 4.

In recent years, laparoscopic surgery has gained popularity as a less invasive alternative to open surgery for RSCDH. The benefits of using laparoscopic techniques, such as mesh repair and suturing, are reduced postoperative pain, shorter hospital stays, and faster recovery [19, 20].

Table 4. Risk factors of poor outcome in congenital diaphragmatic hernia

Risk factors
• Right-sided CDH
• Low LHR value
• Concomitant cardiac defects
• Concomitant extracardiac defects
• Low diameter of ascending aorta
• Low heart area/chest area value
• Preterm delivery

CDH – congenital diaphragmatic hernia, LHR – lung-to-head ratio.

Conflict of interest

The authors declare no conflict of interest.

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