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Associated intraabdominal malformations of right-sided congenital diaphragmatic hernia: A rare anomaly and review

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ABSTRACT

There is scarce literature on associated anomalies of right-sided congenital diaphragmatic hernias (CDH). The purpose of this study was to expand the presentation of a unique clinical experience by a literature review. Only six articles of a right-sided CDH linked to non-cardiac anomalies with complicated diagnostic and treatment scenarios were found related to ours. To the best of our knowledge, it is the first presented case of a right-sided CDH with multiple intestinal atresias and an intrathoracic right kidney. This case alerts physicians to take all examination details into account to avoid delaying CDH diagnosis. Most importantly, it gives valuable insight into possible associated anomalies of right-sided congenital diaphragmatic hernias and may benefit future embryological or genetic studies.

1. Introduction

Congenital diaphragmatic hernia (CDH) is described as a defect in the diaphragm that allows abdominal organs to herniate into the thoracic cavity. CDH is a rare anomaly with an occurrence rate ranging from one to five in 10,000 live births [1]. According to existing literature, right-sided diaphragmatic hernias are less common (15%) than left-sided (85%) and carry a higher risk of mortality and morbidity [2]. In 40–50% of cases, CDH is associated with other congenital anomalies such as central nervous system defects, cardiac and genitourinary pathologies. However, associated non-cardiac congenital malformations of right-sided CDH are underrepresented in literature. Its coexistence with intestinal atresia is extremely rare, with a single case of a right-sided CDH and duodenal atresia reported in literature [3–5]. Coexistence with intrathoracic kidney was reported but the combination of both duodenal and jejunal atresias, with intrathoracic renal ectopia is unique. The aim of this study was to analyse complicated diagnostic and therapeutic management and to conduct a literature review.

2. Methods

Informed consent from patient's parents was obtained. Demographic characteristics, clinical history, imaging data and management were assessed from medical documents and electronic data files. The literature search was made using the electronic National Institute of Health database PubMed.gov. For the search terms "Right-sided diaphragmatic hernia AND duodenal atresia AND jejunal atresia AND intrathoracic renal ectopia" were used, however, no results were found. Further search was performed in order to select

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any linked papers using search terms "Right-sided diaphragmatic hernia AND congenital anomaly". From 273 detected articles, six papers relevant to our case were selected. Experience of other centers presented in Table 1 was compared to our clinical experience.

3. Case description

A female neonate weighing 1600 g with Apgar scores of eight at the first and the fifth minutes was born through a standard vaginal delivery at 30 weeks of gestation and referred to our tertiary level neonatal intensive care unit (NICU). Her prenatal ultrasound showed signs of duodenal atresia at the 26th gestational week with no other signs of congenital malformations. The baby presented with signs of proximal bowel obstruction and respiratory distress, due to which continuous positive airway pressure (CPAP) was initiated. An abdominal ultrasound revealed an aperistaltic, dilated duodenum, filled with liquid content, and distally empty intestinal loops, consistent with duodenal atresia findings. A renal ultrasound identified a single left kidney with suspected right kidney agenesis. Chest X-ray showed an unclear opacification of lower segments of the right lung, reduced lung aeration, and distended stomach (Fig. 1). No pleural effusion or pathological changes in diaphragm integrity were observed by ultrasound of pleura and lungs or sus-

Table 1
Previously reported cases of right-sided CDH and associated anomalies.

Study (year of publication)	Associated anomaly	Existence of cardiac anomaly	Birth weight, g	Time of CDH diagnosis (prenatally/postnatally)	Outcome
Castle et al. (2011)	VACTERL	Yes	3010	Postnatally	Survived
Chen et al. (2013)	VACTERL	Yes	2830	Postnatally	Survived
Olenik et al. (2014)	Hepatopulmonary fusion	No	2885	Prenatally	Survived
Juricic et al. (2016)	Intrathoracic kidney	No	3905	Prenatally	Survived
Takezoe et al. (2017)	Intrathoracic kidney; hepatopulmonary fusion	No	2780	Prenatally	Survived
Almaramhy et al. (2018)	Hepatopulmonary fusion	No	Not presented	Postnatally	Died on the 3rd postoperative day

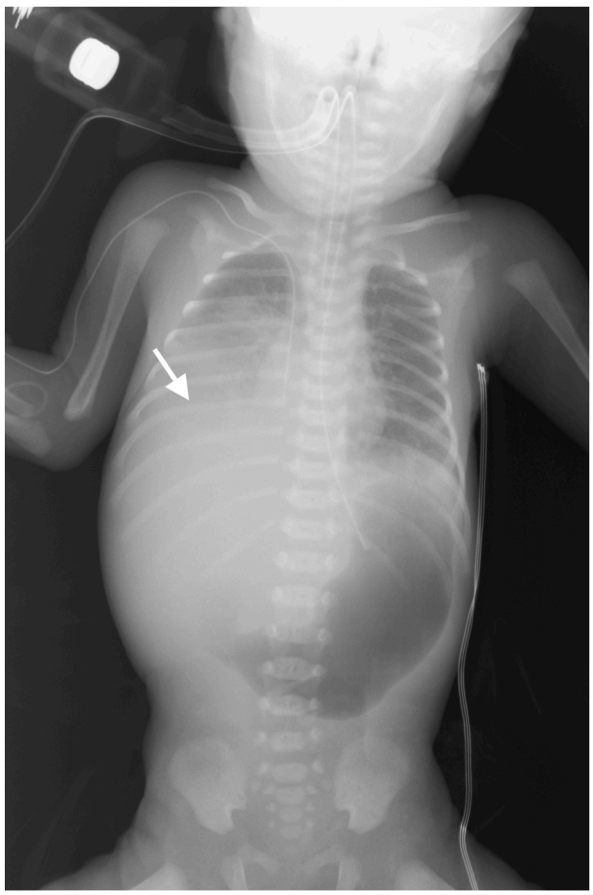

Fig. 1. 2nd day chest x-ray. Undefined opacification of lower right lung segments and reduced lung aeration (white arrow).

pected on chest x-rays. Right lung atelectasis was believed to be the cause of the impaired lung function, and the baby was subsequently intubated.

A laparotomy was done for duodenal atresia repair. During the operation, a segment of jejunum was observed in adhesions. Having dissected it, a 3cm jejunal atretic segment with recanalization on its medial side was observed (Fig. 2). Duodenal atresia was repaired by a Kimura duodenoduodenostomy, while the atretic nonfunctioning jejunal segment was excised, and a feeding tube was easily passed through the recanalized jejunum. The surgeons did not identify a diaphragmatic hernia during the procedure since the liver was lying in its proper position covering the diaphragmatic defect.

Due to deteriorating respiratory distress postoperatively, an ultrasound of the pleura and lungs was carried out. It revealed an additional pulmonary mass with accessory arterial supply from the aorta, implying a possible pulmonary sequestration. In addition, the ultrasound showed an abnormally high positioned right kidney. However, subsequent ultrasonographic examination alongside renal ectopia suggested intrathoracic anomalous liver tissue and likely heterogeneity of the right diaphragm (Fig. 3). It was the first time the newborn was suspected of having a diaphragmatic hernia. A chest computed tomography (CT) (Fig. 4) was obtained to differentiate between a pulmonary sequestration and possible hepatopulmonary fusion, however it did not clarify the final diagnosis. Subsequently, on the fourth postoperative day, a diagnostic right thoracoscopy (Fig. 5) was performed, and a 4cm right-sided diaphragmatic hernia with herniation of the right kidney and upper liver edge was finally confirmed. On account of the high kidney position, conversion to laparotomy was chosen, and, having returned abdominal organs to the abdominal cavity, the diaphragmatic hernia was repaired with single sutures. The second postoperative period was uncomplicated.

4. Discussion

To the best of our knowledge, there has only been one case of a right-sided CDH associated with duodenal atresia reported in literature [4]. Our presented case is the first described right-sided CDH with multiple intestinal atresias and a right intrathoracic kidney. We were able to find very few clinical cases of various non-cardiac associated anomalies of a right-sided CDH (Table 1).

Congenital diaphragmatic hernia is a rare clinical finding, and its etiology is not fully understood. It has been proposed that the development of CDH might be influenced by nutritional deficiencies, environmental exposure, and genetic factors [6]. One of the etiological theories suggests that low vitamin A intake might be the driving mechanism of abnormal diaphragm embryogenesis through impaired retinoid signaling. In 2021, Rocke et al. published a study in which, for the first time, a correlation between inadequate maternal vitamin A intake and susceptibility to teratogen-induced CDH was demonstrated in a rat model [7]. According to the latest literature, about 30% of CDH cases in neonates are caused by genetic factors, such as mutations of the WT1 transcription factor (WT1), chick ovalbumin upstream promoter transcription factor II (COUP-TFII), GLI-Kruppel family member 2 (GLI2), and others [8]. CDH has been linked with autosomal recessive, autosomal dominant, and X-linked patterns of inheritance. Nevertheless, etiology remains unspecified in more than 70% of all CDH cases [9].

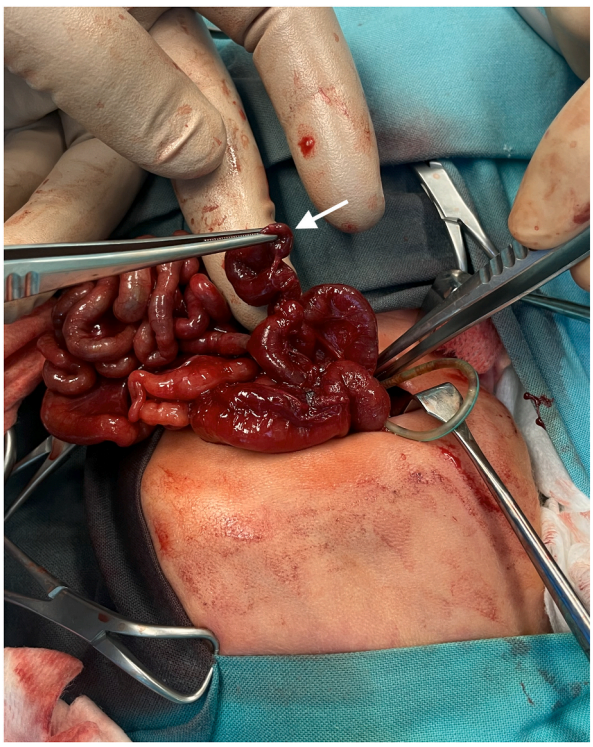

Fig. 2. First surgery. A recognized 3cm jejunal atretic segment (white arrow) during the first surgery.

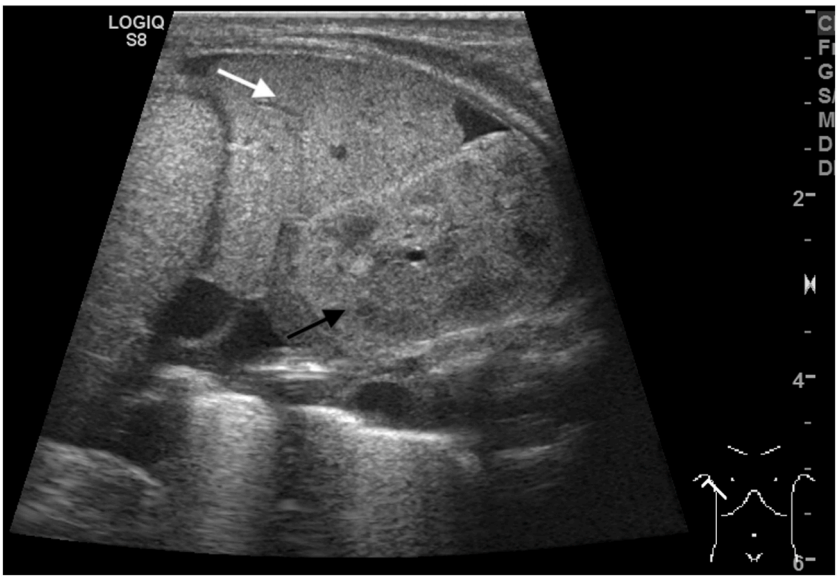

Fig. 3. Post-operative thoracic ultrasonography. Intrathoracic renal ectopia (black arrow) and possible intrathoracic liver tissue (white arrow).

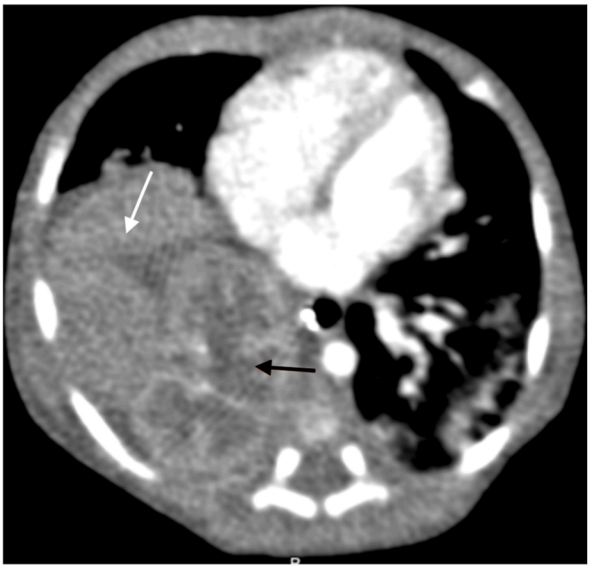

Fig. 4. Post-operative chest CT. Intrathoracic renal ectopia (black arrow) and possible intrathoracic liver tissue (white arrow).

Furthermore, some authors have suggested that right and left-sided CDH may be ethiopathogenetically different and that the type of associated nondiaphragmatic malformations may be linked to the side of the diaphragmatic defect [10,11]. R. Grizjel has disputed this theory, however there is still a significant lack of literature regarding concomitant malformations of right-sided CDH in order to make complete conclusions [12]. Reporting on and analyzing associated congenital malformations may help decipher the embryonic development of CDH and conclude if right and left CDH are two distinct entities.

There is varied literature on antenatal diagnosis of the right-sided CDH. According to Jani et al., right-sided CDH is challenging to diagnose prenatally by ultrasound in cases where the liver is the only organ that has herniated because of its similar echogenicity to that of lungs [5]. Although, in their study Hedrick et al. succeeded in diagnosing right-sided CDH prenatally in 81,5% of all cases, current literature implies that in contrast to right-sided, left-sided congenital diaphragmatic hernias are easier to diagnose antenatally because they may appear as heterogeneous masses in the thoracic cavity [6,13,14].

If a diaphragmatic hernia remains unidentified before birth, there are certain key points to follow in order to diagnose it postnatally. CDH might cause diminished breath sounds on the ipsilateral side of the hernia, a shift of heart sounds contralaterally, and a scaphoid abdomen. Visible bowel gas above the diaphragm and mediastinal shift on chest x-ray confirm the presence of diaphragmatic hernia [15–18]. Sometimes a right-sided CDH can be mistaken for pneumonia or lung atelectasis on a chest X-ray as reported in the case described by Duan et al. and as was our experience [19]. It is important to note that in the presence of an unidentified in-

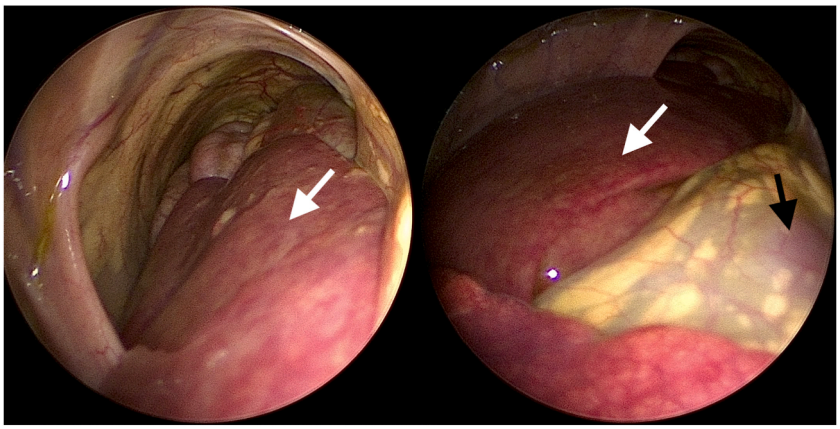

Fig. 5. Diagnostic thoracoscopy. Right-sided diaphragmatic hernia with herniation of the right kidney (black arrow) and upper liver edge (white arrow).

trathoracic mass and the absence of the kidney ipsilaterally, CDH should always be suspected [20,21]. The difficult diagnostic process that we faced adds up to existing literature stating that right-sided CDH often requires complex examination [6,14].

In our clinical case, duodenal atresia was diagnosed on the 26th gestational week using transabdominal ultrasound, whereas CDH was not suspected before birth. In the case described by Castle et al., duodenal atresia was also diagnosed prenatally, while CDH was confirmed after the first neonatal chest X-ray. Early postnatal CDH diagnosis was aided by the fact that a dilated duodenum could be seen in the thoracic cavity as opposed to our clinical case experience [4]. In our presented tricky clinical scenario, the original chest x-ray did not raise any suspicion of herniated viscera. Retrospectively, we believe that a few facts complicated the diagnosis of CDH in this clinical case. Firstly, the liver was covering the diaphragmatic defect, so the abdominal organs could not pass through it into the chest. Most likely, the right kidney had already been herniated into the thoracic cavity when the first chest x-ray was performed, however as there were no clear margins of the opacity in the right thorax, a solid mass was not considered. Owing to the little amount of herniated content and low intrabdominal pressure caused by duodenal atresia, lung development of the neonate was not strongly disrupted, leading to atypical CDH symptoms. After managing duodenal atresia, the bowel began to function, and, under increased intraabdominal pressure, the upper liver edge protruded into the thoracic cavity, causing respiratory distress and subsequent right-sided CDH diagnosis.

5. Conclusion

The coexistence of a right-sided congenital diaphragmatic hernia and multiple intestinal atresias, and intrathoracic renal ectopia in the same individual is an exceptional clinical finding. Diagnosing this type of hernia might be challenging. All examination details must be taken into account and considered thoroughly in order to avoid complications that may be fatal. In the absence of a right kidney and ipsilateral lung opacity, right-sided CDH should be considered and operative management should be planned accordingly in order to avoid possible complications. This rare case can provide further insight into the pathophysiology and associated anomalies of right-sided congenital diaphragmatic hernias and benefit future embryology studies.

Patient consent

Informed consent from patient's parents was obtained.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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