Ref: Ro J Med Pract. 2022;17(4)
DOI: 10.37897/RJMP.2022.4.4

Congenital diaphragmatic hernia with pneumothorax, a challenge for the neonatologist on call

Ioana-Luciana CONSTANTIN[1,2,3], Diana-Sanziana ARNAUTU[1], Delia STRATONE[1,2], Elena ROTARU[1], Adelina TUTA[3], Anda AVASILCAI[4], Vlad DIMA[5], Livica FRATIMAN[1,2], Irina FRANCIUC[1,2]

[1]"Ovidius" University, Faculty of Medicine, Constanta, Romania
[2]"Sf. Andrei" County Clinical Emergency Hospital, Constanta, Romania
[3]"Iuliu Hatieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania
[4]"Cuza Voda" Clinical Hospital of Obstetrics and Gynecology Iasi, Romania
[5]Filantropia Clinical Hospital, Bucharest, Romania

Abstract

Objectives. *Congenital diaphragmatic hernia (CDH) represents a developmental defect of the diaphragm, which allows the protrusion of the abdominal viscera into the thoracic cavity. In view of the fact that herniation occurs during a critical period of lung development, the pathological effect is pulmonary hypoplasia in different degrees, usually more severe on the ipsilateral side of the hernia, and also may be present contralateral if the mediastinum is bulged, compressing the lung.*

Material and methods. *We present the case of a patient hospitalized in the Neonatology Department of "St. Andrew" County Emergency Clinical Hospital of Constanta, diagnosed with left CDH at birth.*

Outcomes. *Full-term male newborn, gestational age (GA) 39 weeks, birth weight (BW) 3300g, Apgar Score 6. The 35-year-old mother, Gravida-VI, Para-III, has a pregnancy with inadequate prenatal care, with no structural abnormalities of the fetus detected by the 3rd-semester ultrasound. At birth, the newborn needed neonatal resuscitation, initially with positive pressure ventilation (on mask and bag) and after that, intubated and mechanical ventilated. Chest X-Ray showed a left diaphragmatic hernia, and the pediatric surgery team was called for further therapeutic management. Approximately 48 hours postoperatively, the chest X-Ray identified right upper lobe pneumothorax, with complete remission in 24 hours, under mechanical ventilation.*

Conclusions. *CDH represents a condition with a challenging diagnosis and management. In the best cases, newborns have a very good clinical outcome with neonatal care and surgical treatment after birth. The management of infants with congenital diaphragmatic hernia requires the services of an interprofessional team. After the diagnosis in the antenatal period, parents should be allowed to discuss with a team, including maternal-fetal medicine, pediatric surgery, neonatology, and social work as appropriate. Genetic evaluation and counseling are recommended to identify risks in future pregnancies. Following the repair in the postnatal period, a standardized and interdisciplinary follow-up to provide surveillance, screening, and clinical care is recommended to improve outcomes.*

Keywords: congenital diaphragmatic hernia, pulmonary hypoplasia, pneumothorax

Corresponding authors:
Constantin Ioana Luciana
E-mail: constantin.ioana1996@gmail.com

Article History:
Received: 5 December 2022
Accepted: 9 December 2022

INTRODUCTION

CDH refers to a developmental defect of the diaphragm that is evident at birth in most individuals. It is an uncommon disorder characterized by the incomplete genesis of the diaphragm resulting in absence, deficiency, or eventration resulting in elevation of a portion of the diaphragm that is thinned as a result of incomplete muscularization [1].

The protrusion of abdominal contents characterizes CDH into the thoracic cavity affecting the normal development of the lungs, resulting in variable degrees of pulmonary hypoplasia (the main complication of CDH) associated with a decrease in cross-sectional area of the pulmonary vessels and the alteration of the surfactant system. The incidence based on the available literature ranges from 0,8-5/10.000 birth with variability across the population. The male/female ratio is to some degree but not much in favor of the masculine gender. Nevertheless, Morgagni's hernias have no clear evidence of gender association. The high mortality and morbidity rates associated with CDH are directly related to the severity of cardiopulmonary pathophysiology. Although the etiology remains unknown, it has a polygenic origin in approximately one-third of cases. The most common syndromic causes of CDH are Pallister-Killian syndrome, Fryns syndrome, Wolf-Hirschhorn syndrome, Cornelia de Lange syndrome, craniofrontal nasal syndrome, Donnai-Barrow syndrome, Simpson-Golabi-Behmel syndrome, CHARGE syndrome, Denys-Drash syndrome, Apert syndrome, Beckwith-Wiedemann syndrome, and 22q11.2 deletion syndrome [2-3].

The survival rate of patients with a congenital diaphragmatic hernia after surgery is between 70-92%. There are three basic types of CHD: the posterolateral "Bochdalek" hernia, the anterior "Morgagni" hernia, and the hiatus hernia.

The pathological effect is pulmonary hypoplasia in different degrees, representing the incomplete development of the lung tissue, with a reduced number of cells, blood vessels, and alveoli affecting the gas exchange.

Immediate postnatal intubation avoiding mask ventilation should be carried out, and small volume, high frequency, and reduced peak pressure mechanical ventilation should be started [3-4].

CASE PRESENTATION

After neonatal reanimation in the delivery room, the newborn was admitted to NICU on continuous mandatory ventilation (CMV), with a 21% FiO2 and prophylactic antibiotic therapy. The Chest-X ray showed: a left diaphragmatic hernia with the intestinal loops occupying almost the entire left side of the chest and the contralateral deviation of the mediastinum (Figure 1).

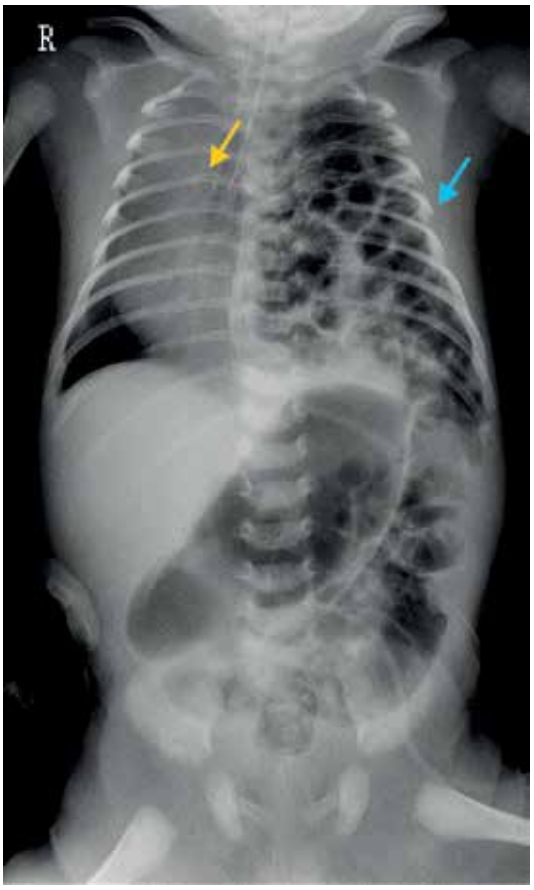


FIGURE 1. *Abdominal Chest X-ray showing: Left diaphragmatic hernia with the significant ascent of the intestinal loops, occupying almost all the left side of the chest (blue arrow) and contralateral deviation of the mediastinum (yellow arrow)*

Before surgery, the newborn was stable on CMV mode with a 40% FiO2 and total parenteral nutrition (TPN). At 36 hours of life, the surgery is successfully performed, the intestinal loops are carefully placed in the abdominal cavity, and the left lung base seems normally colored through the diaphragmatic defect and easily participates in respiratory movements. There are no intestinal congenital abnormalities.

After surgery, the baby is readmitted to NICU, and intubated in CMV mode. The chest X-ray reveals the right upper lobe pneumothorax (Figure 2).

We increased the FiO2 to 100% for one day with complete pneumothorax remission (Figure 3) then we returned to 21% FIO2 on mandatory ventilation for another 2 days.

We noticed that the baby was breathing by himself because the endotracheal probe was no longer in position. From that moment he no longer needed respiratory support, and remains on TPN because of the continuous biliary drainage on the nasogastric tube.

On the 5th day, we performed abdominal contrast radiography (Figure 4 and Figure 5) showing a quasi-complete gastric and intestinal evacuation.

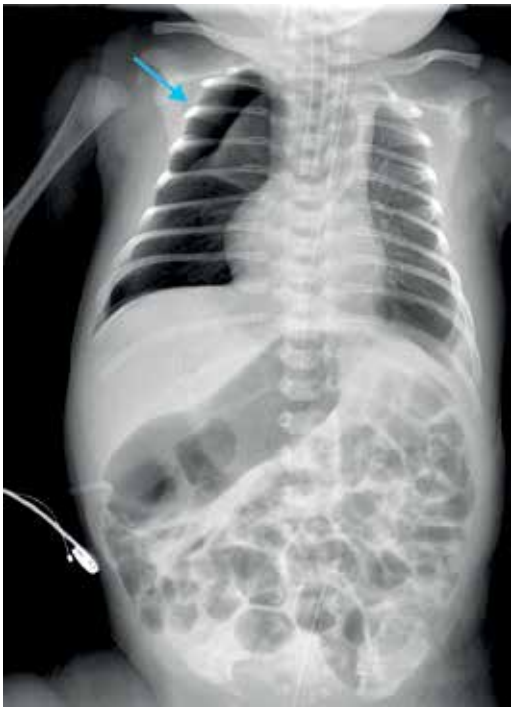


FIGURE 2. *Post-operative, chest X-ray showing: right upper lobe pneumothorax (blue arrow)*

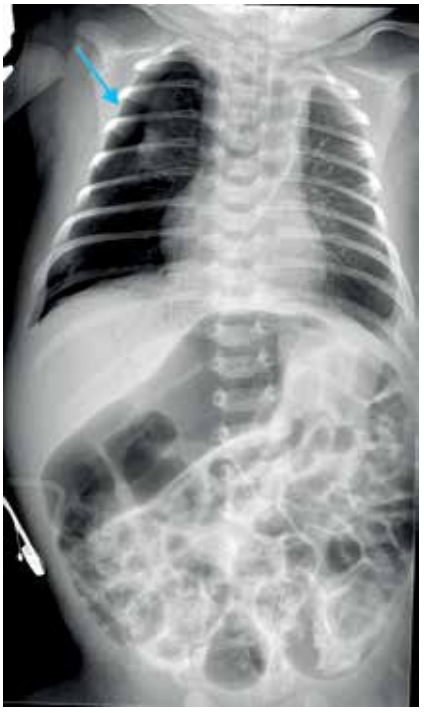
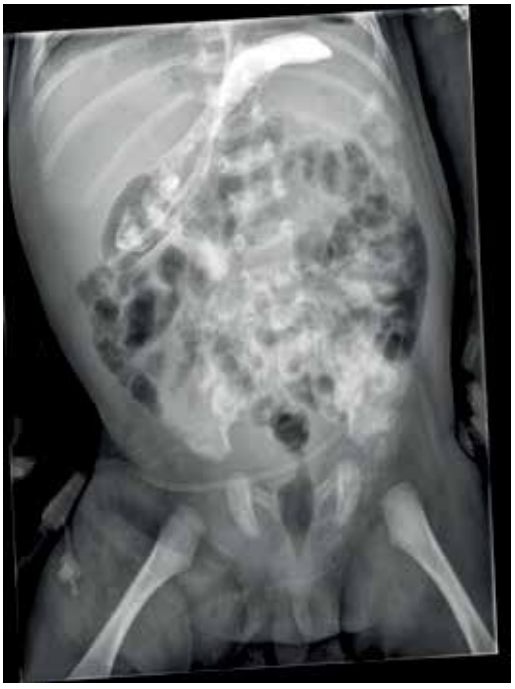


FIGURE 3. *Right upper lobe pneumothorax (blue arrow) that got absorbed by itself under VM-CMV with 100% FiO2 in approximately 24 hours*



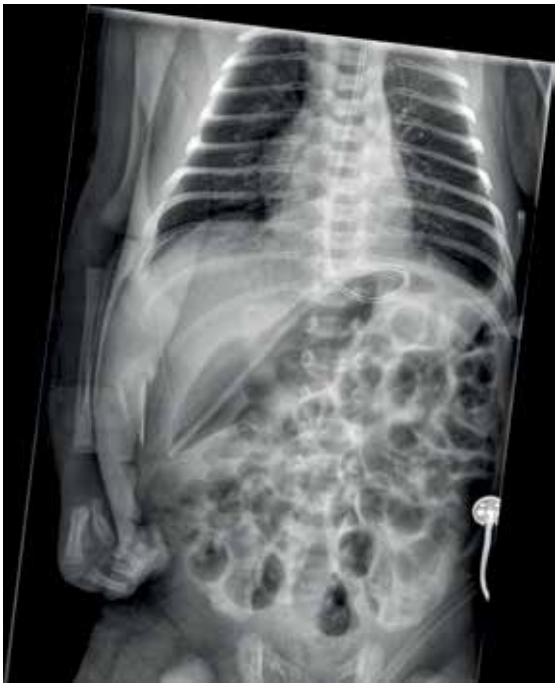


FIGURE 4-5. *X-ray showing a normal digestive tract*

OTHER INVESTIGATIONS

- Cranial ultrasonography – within normal limits.
- Echocardiography – within normal limits, heart malformations are excluded.
- Ophthalmological examination-completely vascularized retina.

Despite the postnatal diagnosis, our patient had a favorable outcome after the surgical correction, he was extubated, having a good respiratory adaptation in the atmospheric air, the enteral nutrition was initiated with good digestive tolerance and at 18 days of life (DOF), was discharged home with quasi-normal function for

both respiratory and digestive systems. The early postnatal diagnosis of this case was also due to the fact that we have a mobile radiology service in our NICU that allowed early investigations.

DISCUSSIONS

CDH persists as one of the most difficult perinatology and neonatal surgery situations. The severity of the lesions requires using the complete armamentarium of advanced neonatal care. The rarity of the condition makes setting solid evidence-based management protocols very difficult. The CDH opening can be on the diaphragm's left or right side. The majority of babies have an opening on the left side. The placement of the defect can impact which abdominal organs migrate into the chest. The main difference between the left and right sides is typically the position of the liver. A left-sided CDH allows the abdominal organs: stomach, intestines, and sometimes the liver to move up into the baby's chest. A right-sided CDH typically allows the liver to move into the chest [3-4].

CDH is usually associated with pulmonary hypoplasia, which is not our situation, and that ensures a better outcome for the patient. Pneumothorax is often associated with this pathology. In our case, it developed postoperatively. It usually occurs on the same side as the defect. The contralateral position represents the particularity of our case. There are studies sustaining an increased risk of mortality when it develops preoperatively. It remains a life-threatening complication, even under respiratory management with gentle ventilation. High airway and peak inspiratory pressures (PIP) represent a risk factor for pneumothorax development. Given that the high-frequency ventilation modes become more frequently used, guidelines that address these ventilation settings are very important to avoid complications of mechanical ventilation and outcomes improvement [5].

Studies say that there is an association between vitamin A and CDH. In addition to pulmonary hypoplasia, the pathophysiology of CDH also includes surfactant deficiency. Vitamin A is essential for various aspects of lung development. Epidemiological data suggest that insufficient dietary vitamin A consumption represents a risk factor for developing CDH. The retinoid hypothesis initially proposed by Greer et al. [6] states that abnormal retinoid signaling early in diaphragm development contributes to the etiology of CDH. In this context, retinoid refers to vitamin A and its metabolites, including the potent signaling molecule retinoic acid. We should also keep in mind that there are secondary effects of excessive vitamin A intake, especially at the beginning of the first quarter of pregnancy, cerebral and cardiac congenital malformations, and spontaneous abortion [6-7].

Gas exchange can be affected in infants with CDH with abrupt onset of acidemia and a mixed respiratory and metabolic acidosis in the first hour of life. Early performance of adequate cardiopulmonary support may contribute to more timely stabilization of gas exchange. The therapeutical protocol should include preductal and postductal arterial blood gas measurements [8].

Bedside pulmonary echography is very important in NICU and represents a powerful diagnostic technique, a noninvasive research tool to qualitatively describe several neonatal respiratory disorders. Lung ultrasound is the latest imaging technique: it is a radiation-free, inexpensive, point-of-care tool that the clinician can use at the bedside. In critically ill babies, it can be used to detect life-threatening tension pneumothorax rapidly: an international multicenter study confirmed that it has optimal diagnostic accuracy and is quicker than conventional radiology. Over and above that, it is more accurate than chest trans-illumination (which is also less accurate than conventional radiology). Due to its convenience, lung ultrasound has received increasing attention from neonatal physicians. Nevertheless, clear reference standards and guideline limits are needed to apply this diagnostic modality accurately. POC-LUS is a viable and suitable diagnostic method that can be performed in the NICU at the bedside. It is susceptible and reliable in diagnosing all types of neonatal lung diseases. Furthermore, it has many advantages over Chest-X-Ray and Computed Tomography, such as accuracy, safety, economical, intelligibility, and no risk of adverse effects due to radiation. Therefore, we encourage the use of lung ultrasounds in the NICU. However, the following issues must be considered when learning this imaging modality. In the past several years, lung ultrasound has become one of the most exciting applications in neonatal point-of-care ultrasound (POC-US). Several recent articles have found ultrasound imaging to be an equal, if not more effective diagnostic modality than X-ray. In addition, it is quicker, less expensive, and does not expose patients to the increased risks of ionizing radiation exposure [9-11].

The diagnosis of CHD was challenging, but in our case, the newborn had a very good clinical outcome with neonatal care and surgical treatment after birth. A successful collaboration between neonatology and pediatric surgery represents an essential aspect of therapy. Even though our case was radiologically postnatal diagnosed, it had a favorable evolution after the surgical assessment, at 18 days of life (DOL), and quasi-normal function of both respiratory and digestive systems. The baby was discharged home at 18 DOL. The prenatal diagnosis of the congenital diaphragmatic hernia and the early surgical assessment are the key points for managing this pathology.

Antenatal undiagnosed diaphragmatic hernia can cause the newborn's death. There is a big chance for

the prenatally diagnosed patients to be intubated immediately postnatal. In this way, we can avoid gastric distension and pulmonary compression given by ventilation.

Radiography is much more conclusive than lung ultrasound, but both are extremely useful in diagnosing CDH. The neonatologist should perform bedside lung ultrasound because it is reliable and accurate in differentiating neonatal respiratory conditions, predicting morbidity, and guiding invasive interventions. The mobile radiology machine represents an advantage that we have in our hospital [12-15].

CONCLUSIONS

CDH represents a condition with a challenging diagnosis and management. In the best cases, newborns have a very good clinical outcome with neonatal care and surgical treatment after birth. The management of infants with congenital diaphragmatic hernia requires the services of an inter-professional team. After the diagnosis in the antenatal period, parents should be allowed to discuss the care with a group, including maternal-fetal medicine, pediatric surgery, neonatology, and social work as appropriate. Genetic evaluation and counseling are recommended to identify risks in future pregnancies. Following the repair in the postnatal period, a standardized and interdisciplinary follow-up to provide surveillance, screening, and clinical care is suggested to improve outcomes.

Conflict of interest: none declared
Financial support: none declared

REFERENCES

1. Hansen A, Eichenwald E, Stark A, Martin C. Cloherty and Stark's Manual of Neonatal Care, 9th Edition, Philadelphia: LWW, 2022.
2. Wild KT, Schindewolf E, Hedrick HL, Rintoul NE, Hartman T, Gebb J, Moldenhauer JS, Zackai EH, Krantz ID. The Genomics of Congenital Diaphragmatic Hernia: A 10-Year Retrospective Review. *J Pediatr.* 2022 Sep;248:108-113.
3. Hendrik H, Adzick S. Congenital diaphragmatic hernia in the neonate, Accessed November the 10th, 2022 (https://www.uptodate.com/contents/congenital-diaphragmatic-hernia-in-the-neonate#:~:text=Congenital%20diaphragmatic%20hernia%20(CDH)%20is,to%20be%20incompatible%20with%20life.).
4. Bloss RS, Aranda JV, Beardmore HE. Congenital diaphragmatic hernia: pathophysiology and pharmacologic support. *Surgery.* 1981;89:518.
5. Partridge EA, Peranteau WH, Herkert L, Rendon N, Smith H, Rintoul NE, Flake AW, Adzick NS, Hedrick HL. Right- versus left-sided congenital diaphragmatic hernia: a comparative outcomes analysis. *J Pediatr Surg.* 2016 Jun;51(6):900-2.
6. Greer JJ, Babiuk RP, Thebaud B. Etiology of congenital diaphragmatic hernia: the retinoid hypothesis. *Pediatr. Res.* 2003;53:726–730.
7. Rubalcava N, Norwitz GA, Kim AG, et al. Neonatal pneumothorax in congenital diaphragmatic hernia: Be wary of high ventilatory pressures.*World Journal of Pediatric Surgery* 2022;5:e000341.
8. Greer JJ, Babiuk RP, Thebaud B. Etiology of congenital diaphragmatic hernia: the retinoid hypothesis. *Pediatr Res.* 2003 May;53(5):726-30.
9. Rocke AW, Clarke TG, Dalmer TRA, McCluskey SA, Rivas JFG, Clugston RD. Low maternal vitamin A intake increases the incidence of teratogen induced congenital diaphragmatic hernia in mice. *Pediatr Res.* 2022 Jan;91(1):83-91.
10. Gien J, Palmer C, Liechty K, Kinsella JP. Early Abnormalities in Gas Exchange in Infants with Congenital Diaphragmatic Hernia. *J Pediatr.* 2022 Apr;243:188-192.
11. Raimondi F, Yousef N, Migliaro F, Capasso L, De Luca D. Point-of-care lung ultrasound in neonatology: classification into descriptive and functional applications. *Pediatr Res.* 2021 Sep;90(3):524-531.
12. Ruoss JL, Bazacliu C, Cacho N, De Luca D. Lung Ultrasound in the Neonatal Intensive Care Unit: Does It Impact Clinical Care? *Children* (Basel). 2021 Nov 29;8(12):1098.
13. Liu J, Chi JH, Ren XL, Li J, Chen YJ, Lu ZL, Liu Y, Fu W, Xia RM. Lung ultrasonography to diagnose pneumothorax of the newborn. *Am J Emerg Med.* 2017 Sep;35(9):1298-1302.
14. Long AM, Bunch KJ, Knight M, Kurinczuk JJ, Losty PD; BAPS-CASS. Early population-based outcomes of infants born with congenital diaphragmatic hernia. *Arch Dis Child Fetal Neonatal Ed.* 2018 Nov;103(6):F517-F522.
15. Gomella T, Cunningham M, Eyal FG, Tuttle DJ. Neonatology: Management, Procedures, On Call Problems, Diseases, and Drugs. 7th Ed, New York: McGraw-Hill, 2013.