Bilateral Congenital Diaphragm Agenesis in a Neonate with Hydrocephalus: Case Report

Liqa A Rousan a,*, Bayan Al-Nussair a, Heba Al Qudah a, Ziad A Bataineh b

a Department of diagnostic radiology and nuclear medicine, Jordan University of Science and Technology, PO box 3030, Irbid 22110, Jordan
b Department of general and pediatric surgery, Jordan University of Science and Technology, Irbid, Jordan

ARTICLE INFO

Keywords:
congenital diaphragm hernia
diaphragm agenesis
neonates
hydrocephalus

ABSTRACT

Congenital diaphragmatic hernia (CDH) is one of the most common anomalies in neonates. Diaphragmatic agenesis (DA) is rare and is considered at the extreme spectrum of CDH, it is associated with a higher rate of multiple anomalies.

A male neonate with antenatal diagnosis of CDH and hydrocephalus was born at estimated gestational age of 36 + 4 weeks by emergency Caesarean section due to fetal distress. Chest x-ray showed a common pleuroperitoneal cavity with the liver, spleen, and stomach seen in the thoracic cavity suggesting the diagnosis of bilateral absence of the diaphragm, the neonate died after an hour and a half. DA associated with hydrocephalus has never been previously reported in the literature.

1. Introduction

Congenital diaphragmatic hernia (CDH) describes a broad spectrum of congenital anomalies of the diaphragm ranging from small defects to complete absence. The most extreme variant is agenesis of the diaphragm (DA) and may be unilateral or bilateral. CDH carries a poor prognosis mostly due to pulmonary hypoplasia, although survival rates also depend on the presence or absence of associated major congenital anomalies and chromosomal defects [1,2].

CDH is considered one of the most common congenital anomalies in neonates accounting for 2.4 to 4.1 per 10000 births in Europe [3]. The type of CDH depends on the location of the defect: 1) posterolateral through the foramen of Bochdalek, 2) parasternal in the anterior portion of the diaphragm (Morgagni hernia), 3) septum transversum defects in the central tendon, 4) and hiatal hernia [4]. The posterolateral defect/Bochdalek hernia is the most common type seen in 85-90% of cases [3].

Diaphragmatic defects were described in association with trisomies 18 and 21, vitamin A deficiency, and maternal drug use [5].

Although most of the cases are sporadic, few authors have suggested an autosomal mode of inheritance in cases where siblings were affected with unilateral agenesis of the diaphragm [4].

The most common associated congenital anomaly with CDH is cardiovascular anomalies ranging from septal defects to hypoplastic heart [6].

We present a rare case of bilateral diaphragmatic agenesis with a common pleuroperitoneal cavity associated with hydrocephalus in a male neonate with a sibling with hydrocephalus suggesting a rare syndrome, and we review cases previously presented in the literature.

2. Clinical observation and imaging findings

A male neonate was born at 36 + 4 weeks estimated gestation age with antenatal diagnosis of CDH and hydrocephalus by Caesarean section due to fetal distress. His Apgar scores were 4, 5, 7 at 1, 5 and 10 minutes respectively. The baby was intubated immediately and put on synchronized intermittent mandatory ventilation, and was transferred to the neonatal intensive care unit.

On physical examination, the baby was flaccid, cyanosed, and with a large head, head circumference was 37 cm which is above the 97th percentile (normal 32 ± 3 cm). Antenatally, at the age of 18 weeks estimated gestation age, the atrial width for the lateral ventricles was 10 mm and 11 mm on the right and on the left respectively and remained so until the time of delivery. According to reference ranges of fetal ventricle size by ultrasound provided by Ishola et al, our patient’s measurements were above the 95th percentile [7]. Unfortunately images of

* Corresponding author.
E-mail addresses: larousan@just.edu.jo (L.A. Rousan), bayan.alnussair@gmail.com (B. Al-Nussair), noorjust100@yahoo.com (H.A. Qudah), ziadaba@yahoo.com (Z. A Bataineh).

https://doi.org/10.1016/j.ejro.2020.100270
Received 7 May 2020; Received in revised form 27 August 2020; Accepted 31 August 2020
2352-0477/© 2020 The Author(s). Published by Elsevier Ltd. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).
hydrocephalus were not provided in the records.

No dysmorphic features were noted. Physical chest examination by auscultation revealed decreased air entry; oxygen saturation was less than 70. A chest x-ray was done and showed bilateral pneumothoraces and pneumoperitoneum. Soft tissue densities within both hemithoraces were noted mostly representing the right and left liver lobes and the spleen, the nasogastric tube (NGT) was seen coiled in the intrathoracic stomach with few bowel loops in the left hemithorax as well. Collapsed lungs were noted in the apices bilaterally. The chest x-ray findings were suggestive of a single pleuroperitoneal cavity with absence of both hemidiaphragms (Fig. 1).

The baby became distressed with severe oxygen desaturation and asystole. 10 cycles of cardiopulmonary resuscitation were performed with no success. The baby died at the age of one hour and a half. An autopsy was not performed.

At 18 weeks estimated gestation age amniocentesis was performed, the fetus was normal male karyotype.

The parents were counselled regarding poor prognosis of the fetus, and agreed to no surgical intervention after birth.

The parents were unrelated healthy individuals. An older sibling of our patient suffered from hydrocephalus and was treated by ventriculoperitoneal shunting.

Counselling regarding future pregnancies was performed, the parents wished to conceive again and undergo routine detailed antenatal examinations and amniocentesis. The mother is currently pregnant with her third child at 34 estimated gestational age with a healthy fetus.

3. Discussion

The diaphragm is formed between the 3rd and 8th weeks of gestation by fusion of the: 1) septum transversum which forms the central tendon, 2) the dorsal oesophageal mesentery which gives rise to the median portion of the diaphragm, 3) the pleuroperitoneal membrane which separates the pleural and peritoneal cavities, 4) the muscular portion of the body wall which forms a narrow segment inserting the diaphragm to the ribs and the sternum [4].

CDH is considered one of the most common congenital anomalies in neonates. Unilateral diaphragmatic defects are more common than bilateral defects and are associated with other anomalies in 30-40% in comparison to 70% in bilateral defects. Agensis of the diaphragm is a rare condition with only 10 cases reported in the literature up to 2017 [3]. It was found that 73% of cases with DA have associated malformations where cardiovascular anomalies remaining the commonest as seen in patients with CDH. The survival rate of the affected patients depends largely on the cardiopulmonary function and associated anomalies rather than the size of the defect [2].

The diaphragm muscle precursors originate from the cervical somites, as they migrate to the pleuroperitoneal folds they start to proliferate and differentiate. There are genes that are required for migration of the diaphragm muscle precursors. These genes interact with one another in the embryonal pathway for the development of the brain, heart, lungs, diaphragm, kidneys, and pancreas [3]. Therefore, multiple anomalies are present in patients with CDH and DA.

In a large study conducted by Neville et al reviewing 1833 patients with CDH, 17 had bilateral CDH, 12 had associated anomalies the most common being cardiovascular, one case had Fryns syndrome, and one case had Simpson Golabi Behmel syndrome. None of the patients was reported to have hydrocephalus [6]. In a case study by Fauz et al, 166 cases with CDH were reviewed, 4 of them had hydrocephalus [7]. Nineteen cases reviewed by Baglaj et al showed 2 of the patients who had CDH had associated hydrocephalus and none of the cases with DA had hydrocephalus [1]. In a literature review by Al Adnani et al none of the 10 cases with DA had hydrocephalus [3]. In a study conducted by Radhakrishnan et al on brain magnetic resonance imaging in fetuses with CDH none had hydrocephalus. However, they observed enlargement of the extraaxial spaces [8].

DA is believed to be an extreme form of CDH and not a separate entity according to a study conducted by Baglaj et al [1]. In all the previously reported patients in the literature with unilateral or bilateral DA; none of them had hydrocephalus, although it was observed uncommonly in patients with CDH. Therefore, we believe our case is a rare case, and since our patient had hydrocephalus and so did his sibling; we postulate the possibility of our patient being part of an undiagnosed syndrome such as Fryns syndrome or Bieber syndrome [9].

DA is basically an intraoperative diagnosis and no diagnostic studies or clinical parameters identify the babies during the preoperative period [1]. Therefore, since an autopsy was not performed in our patient, an undiagnosed syndrome remains a suggestion.

Genetic counselling is an important point in cases of congenital malformations to assess in future pregnancies and treatment options.

4. Conclusion

DA is a very rare entity with only few cases reported in the literature. The affected neonates are of a higher risk of associated life threatening anomalies. Therefore, it is essential to do a detailed antenatal scans in fetuses found to have congenital diaphragmatic defects to determine the need for early intervention and for genetic counselling for possible hereditary syndromes and future pregnancy planning for the parents.

Ethical statement

The authors declare that a written consent form was signed by the patient’s parents (the patient was a neonate). The authors declare that the imaging provided with the manuscript is anonymous.

Funding statement

The authors declare that the study was not funded by any party.

The Transparency document associated with this article can be found in the online version.

Transparency document.

References

[1] M. Baglaj, R. Spicer, M. Ashworth, Unilateral agenesis of the diaphragm: A separate entity or an extremely large defect? Pediatr Surg Int 15 (1999) 206–209.
[2] L. Karadeniz, S. Guven, E. Atay, F. Ovali, A. Celayir, Bilateral diaphragmatic defect and associated multiple anomalies, J Chinese Med Assoc 72 (2009) 163–165.

[3] M. Al-Adnani, A. Marnerides, Complete bilateral agenesis of the diaphragm: A case report and literature review, Pediatr Dev Pathol 20 (2017) 66–71.

[4] K. Jasnosz, M. Hermansen, C. Snider, K. Oh, Congenital Complete Absence (Bilateral Agenesis) of the Diaphragm: A Rare Variant of Congenital Diaphragmatic Hernia, Am J Perinatol 11 (1994) 340–343.

[5] H.V. Toriello, J.V. Higgins, A.S. Jones, L.L. Radecki, Pulmonary and diaphragmatic agenesis: report of affected sibs, Am J Med Genet 21 (1985) 87–92.

[6] H.L. Neville, T. Jaksic, J.M. Wilson, P.A. Lally, W.D. Hardin, R.B. Hirschl, et al., Bilateral congenital diaphragmatic hernia, In: Journal of Pediatric Surgery 38 (2003) 522–524.

[7] A. Ishola, C. Asaleye, O. Ayoola, O. Loto, B. Idowu, Reference Ranges of Fetal Cerebral Lateral Ventricle Parameters by Ultrasonography, Rev Bras Ginecol Obstet 38 (2016) 428–435.

[8] D.O. Fauza, J.M. Wilson, Congenital diaphragmatic hernia and associated anomalies: Their incidence, identification, and impact on prognosis, J Pediatr Surg 29 (1994) 1113–1117.

[9] R. Radhakrishnan, S.L. Merhar, P. Burns, B. Zhang, F.Y. Lim, B.M. Kline-Fath, Fetal brain morphometry on prenatal magnetic resonance imaging in congenital diaphragmatic hernia, Pediatr Radiol 49 (2019) 217–223.