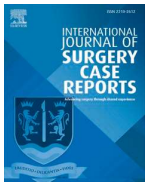
International Journal of Surgery Case Reports 121 (2024) 109969

Contents lists available at ScienceDirect

International Journal of Surgery Case Reports

journal homepage: www.elsevier.com/locate/ijscr



Case report

Incidental discovery of a giant congenital diaphragmatic hernia in an adult: A case report and literature review

Ali Taheriniya [a], Mohammad Reza Maghsoudi [a], Pouria Chaghamirzayi [b,*]

[a] Department of Emergency Medicine, Alborz University of Medical Science, Karaj, Iran
[b] Clinical Research Development Unit of Shahid Madani Hospital, Alborz University of Medical Science, Karaj, Iran

ARTICLE INFO

Keywords:
Congenital diaphragmatic hernias
Adult
Dyspnea

ABSTRACT

Introduction and importance: Congenital diaphragmatic hernia (CDH) is rare, occurring in 1 in 2000 to 4000 live births, and is typically diagnosed in neonates. Bochdalek hernia is the most common type, usually presenting as a left-sided posterolateral defect. Adult presentations of CDH are uncommon and often incidental. This report discusses a young adult with an undiagnosed CDH, emphasizing the importance of clinical awareness.
Case presentation: A 26-year-old man presented with flu-like symptoms and stable vital signs. He reported chronic postprandial shortness of breath that improved with standing. Physical examination revealed decreased breath sounds on the left side. A chest X-ray identified a left diaphragmatic hernia, confirmed by spiral chest computed tomography. Although advised to undergo surgery, the patient opted for discharge against medical advice.
Clinical discussion: Bochdalek hernia, comprising over 95 % of CDH cases, is usually left-sided due to a defect in the pleuroperitoneal membrane. Adults with CDH often present with nonspecific symptoms or the condition is discovered incidentally. Our patient adapted to his symptoms by standing after meals, which provided relief. Surgical intervention is recommended to prevent organ damage, with various techniques available, including open and endoscopic surgery. This case highlights the necessity of clinical vigilance in diagnosing CDH in adults.
Conclusion: Adult congenital diaphragmatic hernia, though rare, requires prompt surgical treatment to prevent organ damage. Recognizing subtle symptoms is crucial for diagnosis. This report contributes to the limited literature on adult-diagnosed CDH, stressing the need for clinical awareness and timely management.

1. Introduction

Congenital diaphragmatic hernia (CDH) is a rare condition. The incidence of diaphragmatic hernia is based on previously performed epidemiological studies of 1 in 2 to 4000 live births. Although there have been reports of diaphragmatic hernia in many newborns, a diaphragmatic hernia report is scarce in adults [1]. There are several variants, the most common being the Bochdalek hernia, which accounts for approximately 90 % of cases. It is characterized by a posterolateral defect, usually occurring on the left side. Other forms include the Morgagni hernia, which is located anteriorly, and central tendon defects. Rarely, patients can reach adulthood undiagnosed and usually present with respiratory and gastrointestinal symptoms [2]. We present a young man with a congenital diaphragmatic hernia. This study has been reported in line with the SCARE criteria [3].

2. Case presentation

We present a 26-year-old man with flu-like symptoms who was referred to the clinic. Written informed consent was obtained from the patient to publish this case report and any accompanying images. His vital signs were stable (No fever, tachypnea, or tachycardia), and his symptoms suggested a common cold. The patient did not have dyspnea and had a dry cough three days before. The patient was lean (Body mass index = 18.2 kg/m^2). In the chest examination, a decrease in breath sounds was evident on the left side of the chest. There is no history of thoracoabdominal trauma. The patient admitted that since childhood, he has felt shortness of breath after every meal, which has improved as long as he stands or walks and usually lasts 45 to 90 min. So, the PA chest X-ray was performed. The left diaphragmatic hernia was seen in the chest X-ray (Fig. 1). For more evaluation, a spiral chest computed tomography scan confirmed the initial diagnosis (Figs. 2 and 3). The patient was introduced for further screening and surgery but discharged

* Corresponding author at: Building Number 25, west Bahar Street, Golshahr Street, Karaj Zip code: 3198636741, Iran.
E-mail address: pouriachaghamirzayi@yahoo.com (P. Chaghamirzayi).

https://doi.org/10.1016/j.ijscr.2024.109969
Received 24 May 2024; Received in revised form 23 June 2024; Accepted 26 June 2024
Available online 27 June 2024

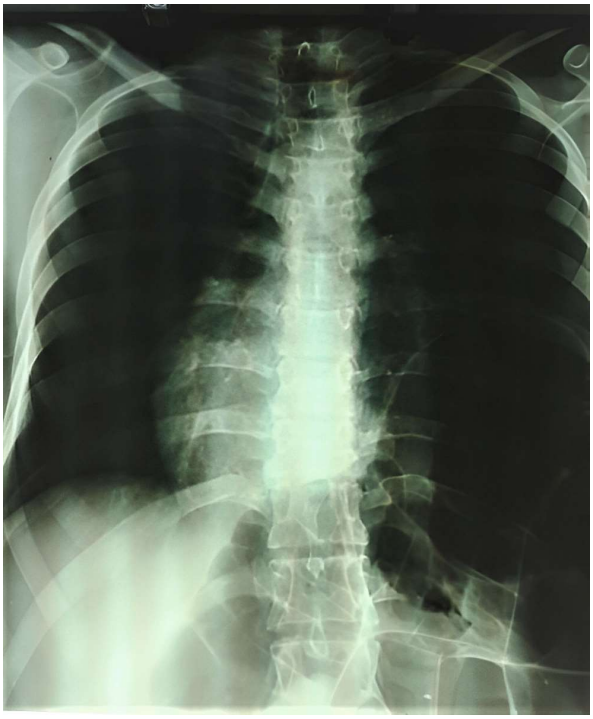


Fig. 1. Chest X-ray: The defect is evident in the left diaphragm.

against medical advice.

3. Discussion

Among the congenital anomalies that are followed by a defect in the closure of the pleuroperitoneal canal posterior to the septum transversum, congenital diaphragmatic hernia (CDH) is the most commonly reported abnormality called Bochdalek defect [4]. More than 95 % of congenital diaphragmatic hernias comprise the Bochdalek hernia, mainly on the left side. On the other hand, the term Bochdalek hernia is a mistake because, in 85 % of cases, an actual hernial sac is not created due to the non-formation of the pleura-peritoneal membrane [5,6]. Late symptoms may include chest discomfort, gastrointestinal symptoms, or hernia strain, leading to a diagnosis. On the other hand, the diaphragmatic hernia may be accidentally seen by accidental chest radiography due to another cause [7,8]. In our case, we accidentally found the diaphragmatic hernia in the chest X-ray. An interesting point about our patient was the adaptation of the patient to dyspnea after eating, which he did by standing up, which helped improve his symptoms. In all adult patients with diaphragmatic hernia diagnosis, to prevent damage to organs, intra-abdominal organs should be repositioned, and defecated diaphragm should be repaired. Various techniques are available for surgical repair of diaphragmatic hernia, including open or less invasive endoscopic surgery [9].

The strength of our report lies in its rarity. It contributes valuable insights into the clinical presentation, diagnostic approach, and management considerations of late-diagnosed CDH. However, our report is limited by its retrospective, single-case nature, which restricts its generalizability to broader patient populations. Additionally, the patient's refusal of surgical intervention precludes insights into long-term outcomes and the efficacy of surgical management. Despite these limitations, our case report provides a foundation for future research to elucidate the epidemiology, clinical features, and optimal management strategies of late-diagnosed CDH.

4. Adult Bochdalek hernia

4.1. Etiology and pathophysiology

Adulthood-diagnosed congenital diaphragmatic hernia (CDH), particularly the Bochdalek hernia, is a serious clinical entity that requires attention due to its implications for patient care and surgical intervention. CDH is a condition characterized by abnormal development of the diaphragm, a part of the lungs, resulting from defects in the septum transversum, pleuroperitoneal folds, body wall, and dorsal mesentery [10–12]. The etiopathogenesis of CDH is unknown, but it was initially believed to be associated with the failure of diaphragmatic closure [10]. The etiology of CDH is unknown in over 70 % of individuals, with some cases showing autosomal recessive, autosomal dominant, and X-linked inheritance patterns [13–15]. Environmental triggers include vitamin A deficiency, exposure to thalidomide or anticonvulsants, mycophenolate mofetil, and allopurinol, which can impair purine biosynthesis [16–18].

4.2. Epidemiology, clinical presentation, and diagnosis

The most common type of CHD is the Bochdalek hernia, a posterolateral defect in the diaphragm. The diagnosis in adults is rare, with less than 200 cases reported [19,20]. Common symptoms of adulthood Bochdalek hernia include shortness of breath, food intolerance, gastroesophageal reflux, nausea, vomiting, abdominal cramping, distension, and abdominal pain [20]. A recent systematic review of Bochdalek hernias in the adult population, including 192 patients, revealed most patients were male (50.5 %) with a mean age of 45.41 $\pm$ 20.26 years. The most common patient symptoms were abdominal pain (62.0 %) and pulmonary symptoms (41.1 %), with the majority of hernias on the left side (70.7 %). A precipitating factor or inciting event contributed to a patient's presentation in 25 % of cases, including obesity, exertion, trauma, pregnancy, laughter, and more. Pregnancy was the most common precipitating factor, while exertion was the most common inciting event [21]. Some cases of congenital diaphragmatic hernias in adults are discovered incidentally, often without prior symptoms.

Various diagnostic techniques are used to evaluate adult patients with Bochdalek hernias. Chest X-ray is the most common initial study, frequently followed by computed tomography (CT) scans, which play a critical role in confirming the diagnosis. Upper gastrointestinal (UGI) or barium studies, and contrast enema are also employed in some cases. Esophagogastroduodenoscopy (EGD) and magnetic resonance imaging (MRI) are less commonly used. Specifically, CT scans are crucial for the final diagnosis of right-sided BH in adulthood, while clinical examination or X-rays rarely lead to a definitive diagnosis. MRI also occasionally contributes to the final diagnosis [22,23]. In adult Bochdalek hernias, the most commonly herniated organs include the colon, small bowel, liver, and kidney. There are also occasional cases where other intra-abdominal organs like the gallbladder or pancreas are involved [20,23].

4.3. Management

Surgical repair is the primary treatment for adult Bochdalek hernias [22]. The surgical options vary, including abdominal approaches such as laparotomy and laparoscopy, as well as thoracic approaches like thoracotomy and thoracoscopy [24–26]. Some cases have been reported that have utilized robotic-assisted techniques [27,28]. The choice between these approaches depends on the patient's condition and the hernia's characteristics. Laparotomy and laparoscopy are common, while robotic-assisted procedures and thoracoabdominal approaches are less frequent [22,23]. Hernia repairs generally involve either direct diaphragmatic sutures or mesh-augmented closures [29–31]. Some cases may combine mesh with sutures or use clips for additional support [32,33]. Modern surgical advancements have made minimally invasive techniques such as thoracoscopic and laparoscopic repairs increasingly

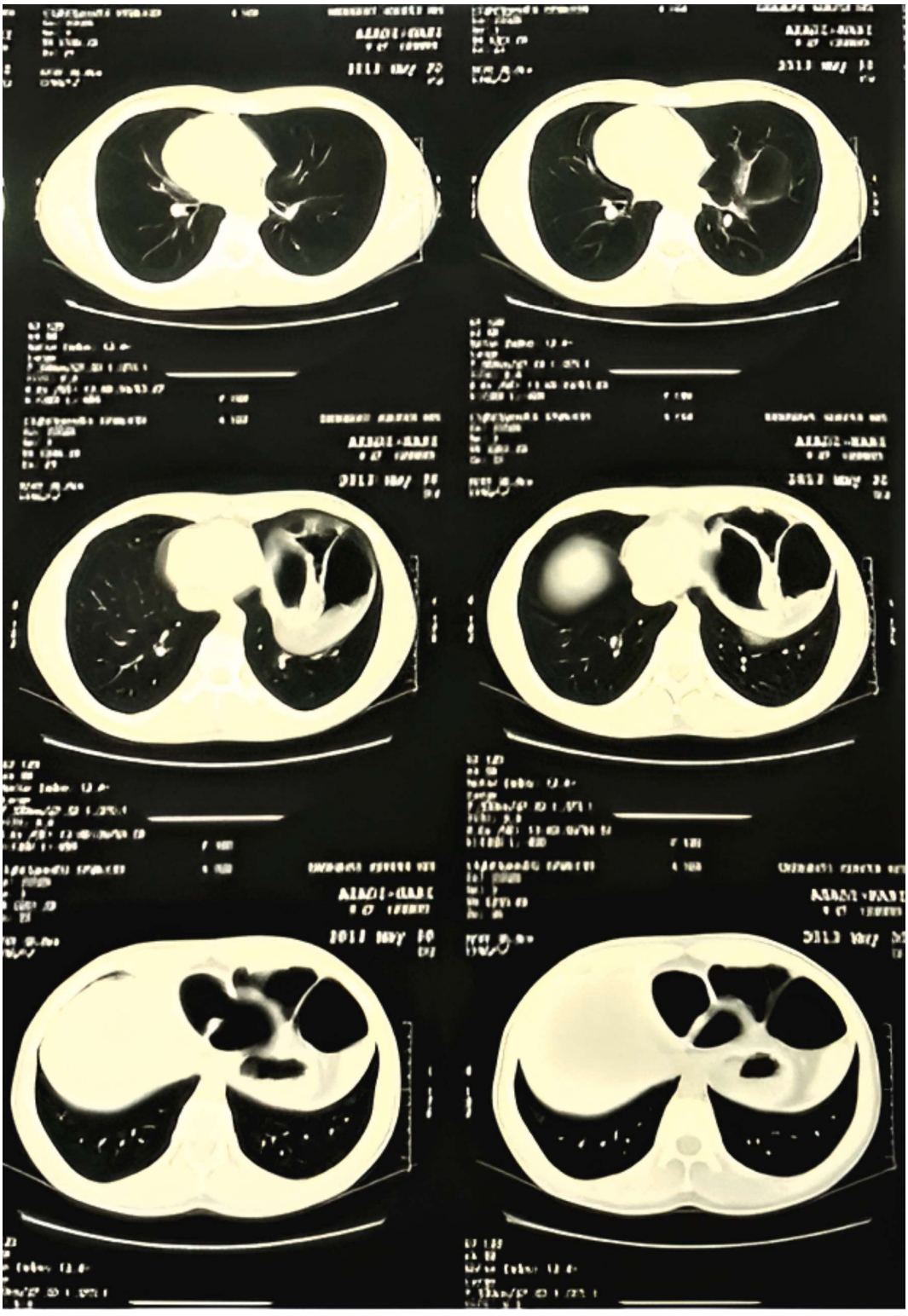


Fig. 2. Chest computed tomography scan (axial view): The presence of abdominal organs on the left side of the thorax.

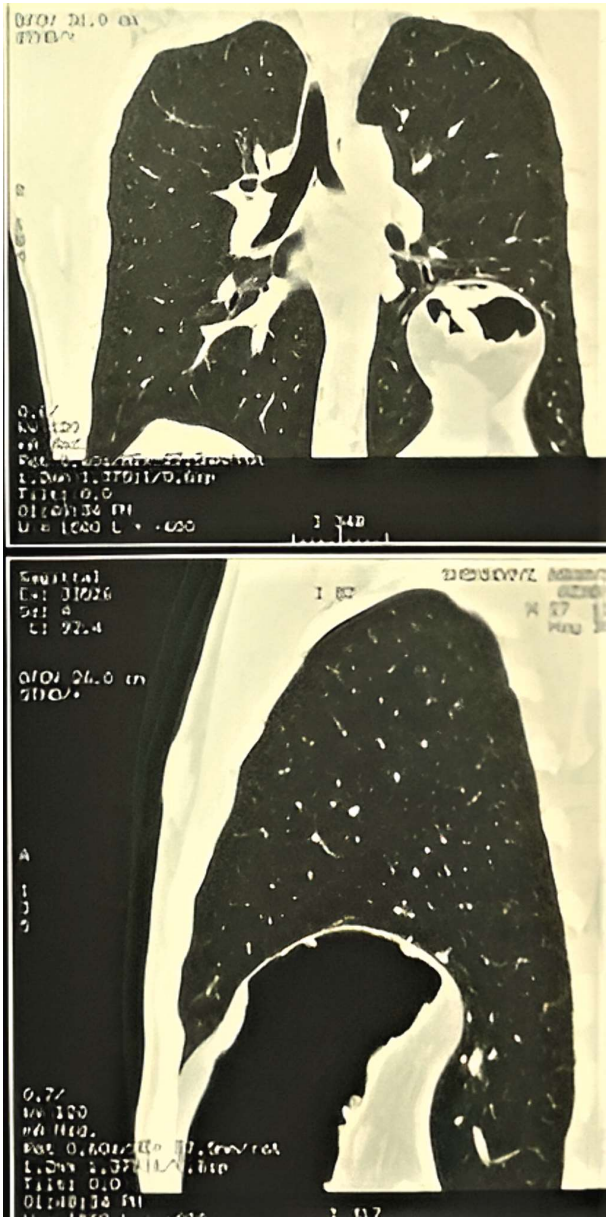


Fig. 3. Chest computed tomography scan (coronal and sagittal views): The presence of abdominal organs on the left side of the thorax.

viable. These techniques offer reduced recovery times and less operative trauma. They are particularly useful in managing incarcerated or strangulated hernias. The laparoscopic approach involves strategic patient positioning to manage complications like pneumothorax during the procedure [19,21,23]. The choice of materials for repair—whether absorbable, non-absorbable, or composite mesh—depends on the specific needs of the repair and the surgeon's preference. Bochdalek hernias typically do not have a hernia sac, unlike other diaphragmatic hernias [21]. When present, management of the hernia sac can vary. Some surgeons recommend excising the sac, while others might use postoperative drains [34,35]. The absence of a hernia sac allows direct communication between the pleural and peritoneal spaces, which requires careful intraoperative handling to prevent pneumothorax [36].

In rare instances, non-surgical management is considered, particularly for patients not fit for surgery or those who refuse it [37,38]. This can involve conservative treatment or specific interventions to manage complications, such as percutaneous nephrostomy or ureteral stenting. These approaches are typically used when patients have mild or no symptoms and when surgery poses significant risks.

4.4. Outcome and complications

Following surgical treatment for Bochdalek hernias, patients generally had favorable outcomes with a significant number showing no hernia-related complications [21,23]. Common issues included urinary complications like hydronephrosis from herniated organs, which were sometimes resolved post-surgery [39]. Thoracic complications such as abscesses and lung infections occasionally require complex interventions [40,41]. Gastrointestinal problems like bowel ischemia and perforation were less reported but required surgical management [42]. Mortality, though low, occurred due to pneumonia, sepsis, and complications from bowel perforation [22]. Patients typically reported improved respiratory function post-repair, despite some experiencing prolonged digestive issues. Overall, the prognosis was positive with rare hernia recurrences reported, underscoring the effectiveness of surgical intervention for Bochdalek hernias [22,23,43].

5. Conclusion

In conclusion, adult congenital diaphragmatic hernia, particularly the Bochdalek hernia, presents as a rare clinical entity with diverse symptomatology and diagnostic challenges. Our case report underscores the significance of prompt recognition through imaging, the variability in clinical presentation, and the critical role of surgical intervention in managing this condition. Despite limitations inherent to single-case retrospective studies and the patient's decision against surgery, our findings highlight the need for a multidisciplinary approach, including advanced imaging modalities and tailored surgical techniques, to optimize outcomes. Future research efforts should focus on refining diagnostic criteria, standardizing treatment protocols, and evaluating long-term outcomes to enhance clinical management strategies for adult CDH.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Ethical approval

All authors and patients agreed to publish this article, and the Alborz University of Medical Science ethics committee approved it (Project number: IR.ABZUMS.REC.1398006). Written informed consent was obtained from the patient to publish this case report and any accompanying images. A copy of the written permission is available for review by the Editor-in-Chief of this journal.

Funding

None.

Author contribution

T.A. interpreted the patient data. P.Ch. M.M. was a significant contributor to writing the manuscript. All authors read and approved the final manuscript.

Guarantor

Pouria Chaghamirzayi.

Conflict of interest statement

The authors declare that they have no competing interests.

Availability of data and materials

All data generated or analyzed during this study are included in this published article (available).

Acknowledgments

We acknowledge the Clinical Research Development Unit of Shahid Madani Hospital, Alborz University of Medical Science, Karaj, Iran, for their support. This research received no funding.

References

[1] Y. Zhou, H. Du, G. Che, Giant congenital diaphragmatic hernia in an adult, J. Cardiothorac. Surg. 9 (2014) 1–3.
[2] H. Manson, Y. Goh, P. Goldsmith, P. Scott, P. Turner, Congenital diaphragmatic hernia causing cardiac arrest in a 30-year-old woman, The Annals of The Royal College of Surgeons of England. 99 (2) (2017) e75–e77.
[3] C. Sohrabi, G. Mathew, N. Maria, A. Kerwan, T. Franchi, R.A. Agha, The SCARE 2023 guideline: updating consensus Surgical CAse REport (SCARE) guidelines, Int. J. Surg. 109 (5) (2023) 1136–1140.
[4] G. Dalencourt, M.R. Katlic, Abdominal compartment syndrome after late repair of Bochdalek hernia, Ann. Thorac. Surg. 82 (2) (2006) 721–722.
[5] W. Yang, S.L. Carmichael, J.A. Harris, G.M. Shaw, Epidemiologic characteristics of congenital diaphragmatic hernia among 2.5 million California births, 1989–1997, Birth Defects Research Part A: Clinical and Molecular Teratology 76 (3) (2006) 170–174.
[6] C.P. Torfs, C.J. Curry, T.F. Bateson, L.H. Honoré, A population-based study of congenital diaphragmatic hernia, Teratology 46 (6) (1992) 555–565.
[7] K.S. Naunheim, Adult presentation of unusual diaphragmatic hernias, Chest Surg. Clin. N. Am. 8 (2) (1998) 359–369.
[8] R.H. JI, Diaphragmatic hernia of Morgagni-Larrey in adults: analysis of 10 cases, Gastroenterol. Hepatol. 26 (9) (2003) 535–540.
[9] F. Moser, F.J. Signorini, P.S. Maldonado, V. Gorodner, A.L. Sivilat, L.R. Obeide, Laparoscopic repair of giant Bochdalek hernia in adults, Journal of Laparoendoscopic & Advanced Surgical Techniques. 26 (11) (2016) 911–915.
[10] G.L. Salas, J.C. Otaño, C.M. Cannizzaro, M.T. Mazzucchelli, G.S. Goldsmit, Congenital diaphragmatic hernia: postnatal predictors of mortality, Arch. Argent. Pediatr. 118 (3) (2020) 173–179.
[11] P. Kosiński, M. Wielgoś, Congenital diaphragmatic hernia: pathogenesis, prenatal diagnosis and management - literature review, Ginekol. Pol. 88 (1) (2017) 24–30.
[12] E. Ersöz Köse, İ. Yalçınkaya, Congenital diaphragmatic hernia, Turk Gogus Kalp Damar Cerrahisi Derg. 32 (Suppl1) (2024) S89–s97.
[13] G. Kardon, K.G. Ackerman, D.J. McCulley, Y. Shen, J. Wynn, L. Shang, et al., Congenital diaphragmatic hernias: from genes to mechanisms to therapies, Dis. Model. Mech. 10 (8) (2017) 955–970.
[14] S.J. Mitchell, T. Cole, D.H. Redford, Congenital diaphragmatic hernia with probable autosomal recessive inheritance in an extended consanguineous Pakistani pedigree, J. Med. Genet. 34 (7) (1997) 601–603.
[15] D.L. Gibbs, H.E. Rice, J.A. Farrell, N.S. Adzick, M.R. Harrison, Familial diaphragmatic agenesis: an autosomal-recessive syndrome with a poor prognosis, J. Pediatr. Surg. 32 (2) (1997) 366–368.
[16] M. Kozenko, D. Grynspan, T. Oluyomi-Obi, D. Sitar, A.M. Elliott, B.N. Chodirker, Potential teratogenic effects of allopurinol: a case report, Am. J. Med. Genet. A 155a(9):2247-52 (2011).
[17] G.M. Enns, V.A. Cox, R.B. Goldstein, D.L. Gibbs, M.R. Harrison, M. Golabi, Congenital diaphragmatic defects and associated syndromes, malformations, and chromosome anomalies: a retrospective study of 60 patients and literature review, Am. J. Med. Genet. 79 (3) (1998) 215–225.
[18] L.W. Beurskens, L.H. Schrijver, D. Tibboel, M.F. Wildhagen, M.F. Knapen, J. Lindemans, et al., Dietary vitamin A intake below the recommended daily intake during pregnancy and the risk of congenital diaphragmatic hernia in the offspring, Birth Defects Res. A Clin. Mol. Teratol. 97 (1) (2013) 60–66.
[19] I. Katsaros, S. Giannopoulos, S. Katelani, M. Vailas, M. Sotiropoulou, D. Papaconstantinou, et al., Bochdalek hernias in the adult population: a systematic review of the literature, ANZ J. Surg. 92 (9) (2022) 2037–2042.
[20] L. Schumacher, S. Gilbert, Congenital diaphragmatic hernia in the adult, Thorac. Surg. Clin. 19 (4) (2009) 469–472.
[21] S.R. Brown, J.D. Horton, E. Trivette, L.J. Hofmann, J.M. Johnson, Bochdalek hernia in the adult: demographics, presentation, and surgical management, Hernia 15 (1) (2011) 23–30.
[22] C.J. Bridges, R.M. Hasson, Congenital hernias in adults: Bochdalek hernias, Thorac. Surg. Clin. 34 (2) (2024) 155–162.
[23] J.P. Ramspott, T. Jäger, M. Lechner, P. Schredl, A. Gabersek, F. Mayer, et al., A systematic review on diagnostics and surgical treatment of adult right-sided Bochdalek hernias and presentation of the current management pathway, Hernia 26 (1) (2022) 47–59.
[24] M. Toda, A. Yamamoto, T. Iwata, Right-sided Bochdalek hernia containing retroperitoneal fat in the elderly: report of a case, Surg. Case Rep. 5 (1) (2019) 81.
[25] N.S. Lau, M. Crawford, C. Sandroussi, Surgical management of symptomatic right-sided Bochdalek hernias in adults: when is a minimally invasive approach appropriate? ANZ J. Surg. 90 (6) (2020) 1075–1079.
[26] C. Palanivelu, M. Rangarajan, G. Maheshkumaar, R. Parthasarathi, Laparoscopic mesh repair of a Bochdalek diaphragmatic hernia with acute gastric volvulus in a pregnant patient, Singap. Med. J. 49 (1) (2008) e26–e28.
[27] A. Jambhekar, S. Robinson, B. Housman, J. Nguyen, K. Gu, V. Nakhamiyayev, Robotic repair of a right-sided Bochdalek hernia: a case report and literature review, J. Robot. Surg. 12 (2) (2018) 351–355.
[28] L.M. Hunter, A.B. Mozer, C.J. Anciano, A.L. Oliver, M.D. Iannettoni, J.E. Speicher, Robotic-assisted thoracoscopic repair of right-sided Bochdalek hernia in adults: a two-case series, Innovations (Phila) 14 (1) (2019) 69–74.
[29] S.J. Baek, J. Kim, S.H. Lee, Hepatothorax due to a right diaphragmatic rupture related to duodenal ulcer perforation, World J. Gastroenterol. 18 (39) (2012) 5649–5652.
[30] E. Laaksonen, S. Silvasti, T. Hakala, Right-sided Bochdalek hernia in an adult: a case report, J. Med. Case Rep. 3 (2009) 9291.
[31] A. Terzi, U. Tedeschi, A. Lonardoni, S. Furia, C. Benato, F. Calabrò, A rare cause of dyspnea in adult: a right Bochdalek's hernia-containing colon, Asian Cardiovasc. Thorac. Ann. 16 (5) (2008) e42–e44.
[32] A.C. Agrafiotis, N. Kotzampassakis, W. Boudaka, Complicated right-sided Bochdalek hernia in an adult, Acta Chir. Belg. 111 (3) (2011) 171–173.
[33] P.A. Shekar, D. Reddy, G. Kochhar, A. Dumra, S. Ks, Herniation of the right renal pelvis through a posterolateral diaphragmatic defect (Bochdalek hernia), Urology 137 (2020) e10–e11.
[34] M. Senkyrik, J. Lata, L. Husová, P. Díte, P. Husa, F. Horálek, et al., Unusual Bochdalek hernia in puerperium, Hepato-gastroenterology 50 (53) (2003) 1449–1451.
[35] W.R. Osebold, R.T. Soper, Congenital posterolateral diaphragmatic hernia past infancy, Am. J. Surg. 131 (6) (1976) 748–754.
[36] K. Takeyama, Y. Nakahara, S. Ando, K. Hasegawa, T. Suzuki, Anesthetic management for repair of adult Bochdalek hernia by laparoscopic surgery, J. Anesth. 19 (2005) 78–80.
[37] Ö. Onuk, T. Taş, A.B. Şentürk, O. Sinanoğlu, M.B. Balcı, O. Çelik, et al., Right-sided Bochdalek hernia with intrathoracic ectopic kidney in an advanced-age adult: a case report, Urol. Int. 93 (3) (2014) 368–370.
[38] P.J. Trivedi, J. Canavan, C. Holloway, A. Slater, S. Travis, An unusual case of dyspnoea in an elderly man, BMJ Case Rep. 2010 (2010).
[39] K. Moro, M. Kawahara, Y. Muneoka, Y. Sato, C. Kitami, S. Makino, et al., Right-sided Bochdalek hernia in an elderly adult: a case report with a review of surgical management, Surg. Case Rep. 3 (1) (2017) 109.
[40] J.M. dos Santos-Netto, C.V. Oliveira, M.G. Sousa, Right-sided Bochdalek hernia in adult associated with cholestatic syndrome: case report, Arq. Bras. Cir. Dig. 28 (4) (2015) 299–301.
[41] M. Watanabe, O. Ishibashi, M. Watanabe, T. Kondo, N. Ohkohchi, Complicated adult right-sided Bochdalek hernia with Chilaiditi's syndrome: a case report, Surg. Case Rep. 1 (1) (2015) 95.
[42] G. Wenzel-Smith, Posterolateral diaphragmatic hernia with small-bowel incarceration in an adult, S. Afr. J. Surg. 51 (2) (2013) 73–74.
[43] S. Katsenos, I. Kokkonouzis, S. Lachanis, K. Psathakis, Right-sided Bochdalek hernia presenting as a solitary pulmonary nodule, Radiol. Case Rep. 3 (2) (2008) 114.