J Afric Neonatal 2025;1: 15 – 18

CASE REPORT

Rukhsar Osman
Naomi A. Mwamanenge
Yaser Abdallah
Karim Manji

A rare case of bilateral congenital pulmonary airway malformation

Received: 14th November 2024
Accepted: 27th December 2024

Rukhsar Osman (✉)
Aga Khan University,
P.O box 2289, Dares Salaam
Email: rukhsarrosman@gmail.com

Naomi A. Mwamanenge,
Yaser Abdallah, Karim Manji
Muhimbili University of Health and Allied Sciences (MUHAS)

Abstract: Congenital Pulmonary Airway Malformation (CPAM) is a rare yet common lung malformation. It can present with respiratory distress at birth or may remain asymptomatic until early childhood. Its incidence in Sub Saharan Africa is not known.
We present a 14-day-old male infant, who was referred with a suspected congenital diaphragmatic hernia based on a chest X-ray. He exhibited severe respiratory distress, and the initial X-ray showed bilateral air-fluid cavities. Intraoperatively, the diaphragm was intact. A subsequent chest computerized tomography (CT) revealed multiple bilateral cystic lesions consistent with CPAM. Despite supportive care, the baby unfortunately succumbed on day 38 of life.
Emphasis should be placed on enhancing antenatal ultrasound skills to detect anomalies early, allowing for timely intervention. Additionally, we advise a CT/MRI scan when a diaphragmatic hernia is suspected on chest X-ray, as this can help rule out other congenital malformations and potentially avoid unnecessary surgery.

Key words: Congenital pulmonary airway malformation, CPAM, congenital diaphragmatic hernia, respiratory distress, pneumonia

Résumé: La malformation congénitale des voies aériennes pulmonaires est une malformation pulmonaire rare. Elle peut se manifester par une détresse respiratoire à la naissance ou rester asymptomatique jusqu'à la petite enfance. Son incidence en Afrique subsaharienne n'est pas connue.
Nous présentons le cas d'un nouveau-né de sexe masculin âgé de 14 jours, qui a été adressé pour une suspicion de hernie diaphragmatique congénitale sur la base d'une radiographie du thorax. Il présentait une détresse respiratoire sévère et la radiographie initiale a montré des cavités bilatérales avec niveaux hydroaériques. En peropératoire, le diaphragme était intact. Une tomodensitométrie thoracique ultérieure a révélé de multiples lésions kystiques bilatérales compatibles avec une malformation congénitale des voies aériennes pulmonaires. Malgré des soins de soutien, le nouveau-né est malheureusement décédé au 38e jour de vie. Il convient de mettre l'accent sur le renforcement des compétences en échographie anténatale afin de détecter précocement les anomalies et de permettre une intervention en temps utile. En outre, nous conseillons d'effectuer une tomodensitométrie/IRM lorsqu'une hernie diaphragmatique est suspectée à la radiographie du thorax, car cela permet d'exclure d'autres malformations congénitales et d'éviter une intervention chirurgicale inutile.

Mots clés : malformation congénitale des voies aériennes pulmonaires, hernie diaphragmatique congénitale, détresse respiratoire, pneumonie

Introduction

Congenital pulmonary airway malformation (CPAM), formerly called congenital cystic adenomatoid malformation of the lung is rare butthe commonest developmental anomaly of the lung accounting for 25% of all congenital lung masses.[1] It consists of hamartomatous or dysplastic lung tissue confined mostly to one lobe of the lung in 95% with the lower lobes being the commonest site, however1-3% cases can be bilateral.[2] The incidence is about 1/25000-35000 live births with an equal left and right-sided presentation and male predominance[3] Its incidence in sub-Saharan Africa is not known.

Pathologically, it is due to adenomatoid proliferation of airway from trachea down to alveoli forming cystic lesions among other normal alveoli. About 60% of cases present in neonatal period,10% in early infancy with respiratory distress and recurrent respiratory infections, and about 15% present in adolescence. Prenatal anomaly screening at 20weeks and beyond with modern ultrasound can detect polyhydramnios due to esophageal compression, and macrocyst (>5cm) or microcysts (<5cm) in the lung appearing as a solid hyperechogenic or cystic lung mass.[4,5]

In Sub Saharan Africa antenatal diagnosis by ultrasound is a challenge due to lack of skilled personnel in sonography in primary health care facilities to pick anomalies. Therefore, this causes a delay in diagnosis and referral of symptomatic fetus to tertiary center for delivery and management of the neonate postnatally. Outcome depends on the presence of fetal hydrops, size of the cyst, pulmonary hypoplasia or mediastinal shift and associated anomalies such as diaphragmatic hernia.[(4)]This report aims at raising awareness on importance of a quality antenatal scan.

Case

We present a term male baby, delivered at 40 weeks of gestation via spontaneous vaginal delivery, with a birth weight of 4.04 kg and APGAR scores of 8 and 9 at the 1st and 5th minutes, respectively. He was born to a para 3, living 3 mother following an uneventful, non-consanguineous pregnancy. Antenatal ultrasounds performed at 20 and 32 weeks of gestation were reported as normal. Her serologies for syphilis, Human Immunodeficiency Virus (HIV), and Hepatitis B were negative, and she denied any drug or substance use.

He developed severe respiratory distress soon after birth, presenting with severe lower chest wall in drawing, grunting, and retractions. He was kept on continuous positive airway pressure (CPAP) support for 8 days with minimal improvement, despite being on maximal settings. Chest x-ray (CXR) done on 8thday of life was reported to have herniated bowel loops bilaterally, and a diagnosis of congenital diaphragmatic hernia was made.

The baby arrived at our facility on the 14th day of life, in significant respiratory distress, and was feeding 80 ml/kg/day of breast milk orally. On examination, the respiratory rate was 77 breaths/min, with intercostal and xiphoid retractions, symmetrical chest expansion, and vesicular breath sounds heard bilaterally. Abdominal examination was unremarkable, with normal contour and bowel sounds present.

The baby was kept on a nasal cannula since the mechanical ventilators available were in use. A nasogastric tube was inserted for decompression, and the baby was kept nil per os. Total parenteral nutrition and ceftriaxone were initiated. An initial diagnosis of sepsis and congenital diaphragmatic hernia was made.

Further workup was done, and a (CXR) revealed bilateral air-fluid cavities of variable sizes. The complete blood count (CBC) showed thrombocytopenia and anemia, C-reactive protein (CRP) was elevated to 58 mg/dl, and blood culture revealed Enterococcus species, which were sensitive to amikacin and meropenem but resistant to ceftriaxone. Serum electrolytes were normal, and the echocardiogram was normal. He was then transfused with packed red blood cells and platelets, and the antibiotics were changed to meropenem.

On day 16 of life, he developed worsening respiratory distress, requiring sedation and mechanical ventilation with SIMV mode. His initial arterial blood gas revealed respiratory acidosis, with a pH of 7.25, PCO2 of 70, and HCO3 of 26. His blood pressure remained normal. A CXR was repeated, which showed multiple cavitating lesions, progressive collapse of the upper right lobe, and worsening patchy infiltrates, as seen in Figure 2. Due to the worsening respiratory condition, surgery was delayed and performed on the 28th day of life. A chevron incision was made, and intraoperative findings revealed an intact diaphragm and intra-abdominal bowels, thereby nullifying the diagnosis of CDH.

A CT chest was subsequently performed on 32nd day of life, revealing bilateral multiple cystic air lesions of varying sizes, affecting all lung lobes, more pronounced on the right. The largest lesion measured 1.65 x 1.52 x 1.32 cm, with consolidation and atelectasis, highly suggestive of bilateral multicystic lung disease with pneumonic changes, as shown in Figure 3. Other differential diagnoses included congenital cystic bronchiectasis.

A cardiothoracic surgeon was consulted and advised supportive care due to the poor prognosis, with minimal success in treatment. Surgical management was considered difficult in our current setting. Therefore, the baby continued on mechanical ventilation despite little improvement, until he succumbed to death on the 38th day of life. A postmortem was not performed due to the family's cultural beliefs

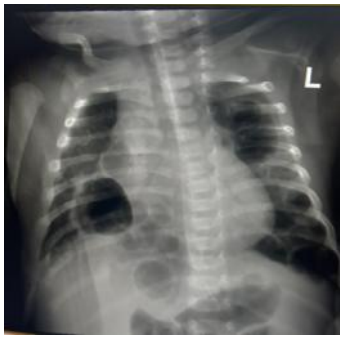

Fig 1: chest x-ray showing bilateral air-filled cavities of variable sizes

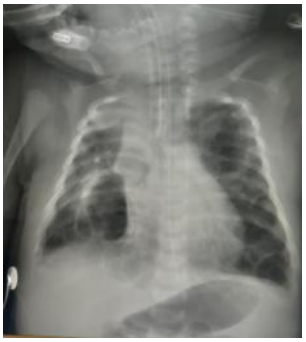

Fig 2: chest x-ray showing multiple cavitating lesions, progressive upper right lobe of lung opacification, collapse and worsening patchy infiltrates

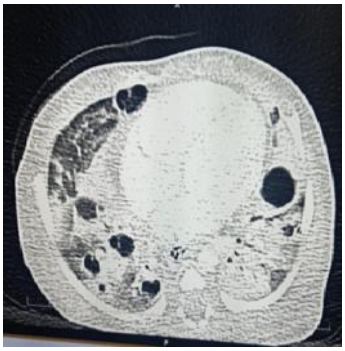

Fig 3: CT chest showing bilateral multiple cystic air lesions of variable size and more in the right lung the largest measuring 1.65 x1.52x1.32cm with consolidation and atelectasis pointing to a more likely diagnosis of bilateral multicystic lung disease with pneumonic changes, atelectasis and consolidation

Discussion

CPAM is ahamartomatous proliferation of terminal bronchioles caused by arrested alveolar development.[6]
In 2002 a modified Stocker classification was made consisting of five types of CPAM from type 0to type 4.[7] The cysts can communicate with the normal tracheobronchial tree which can lead to air trapping causing cystic dilatation of the bronchiole and subsequent respiratory distress, and the CPAM portions does not participate in normal gas exchange. The blood supply and drainage is from pulmonary system.[8] It is an uncommon, and isolated phenomenon, rarely associated with chromosomal defects.[9]

With proper skills in antenatal ultrasound, CPAM can be detected in utero.[10] However, due to lack of resources and skilled sonographers at primary health care facilities, prenatal detection of congenital anomalies in our country is not possible and therefore referral to tertiary centers for delivery is delayed.
Postnatal chest CT confirms its presence in 86% of cases, and associated anomalies can be picked through screening by abdominal ultrasound or echocardiogram.[5]
Differential diagnosis of cystic lesions other than CDH should be considered whenever a CXR is suggestive.[11]
In most of our primary health facilities, CXR is often not available and therefore the neonate would be referred for diagnosis of the ongoing disease. This also in turn delays the neonate to get proper postnatal management.
Clinical presentation can begin in utero with symptomatic fetuses showing cystic lung lesions of varying sizes. These cysts can restrict lung growth, cause mediastinal shift, cardiovascular compromise, vena cava compression leading to non-immune hydrops, or result in pulmonary hypoplasia. Symptomatic neonates may present with respiratory distress, with 40% showing acute progressive symptoms such as tachypnea, retractions, grunting, and sometimes cyanosis.[12] These symptoms are similar to other lung malformations, requiring thorough examination for accurate diagnosis and management.[11] Our neonate's presentation resembled CDH, leading to the initial surgical diagnosis. Some infants are asymptomatic at birth and are diagnosed in early childhood due to recurrent chest infections.
Intervention for fetuses with hydrops can help alleviate symptoms and reduce the risk of pulmonary hypoplasia, although this is not performed in our country due to lack of resources and expertise. Steroids may also be given for large CPAM. Postnatal management includes mechanical ventilation for moderate to severe respiratory distress and resection of the affected lobe to prevent recurrent infections, air leaks, and neoplasm transformation. For asymptomatic patients, surgery can be delayed, though optimal timing is unknown.[13] The challenge in our case was the bilateral nature of the cysts affecting all lobes, making surgical resection difficult and futile.
Complications can be grouped into early and late categories. Early complications include sepsis, air leak, and bronchopleural fistula, while late complications include respiratory symptoms, such as pneumonia.[14] This baby developed pneumonia, which worsened his respiratory status, and the respiratory support provided was ineffective.
Prognosis depends on the neonate's symptoms and associated anomalies. Unilateral (macrocyst) type 1, in the absence of hydrops, mediastinal shift, and polyhydramnios, has a good prognosis. However, large cysts causing fetal hydrops and mediastinal shifts, along with associated anomalies and bilateral cysts, have a poor prognosis, as in the case we present, due to difficulties in surgical resection of the lobes.[15] There is also a risk of malignant transformation in type 1 and type 4 CPAM, leading to bronchioalveolar carcinoma (BAC) or pleuropulmonary blastoma (PPB), respectively.[16]

Conclusion

CPAM is a rare entity and should be suspected in an infant presenting with features suggestive of congenital diaphragmatic hernia clinically and radiologically. In our case emphasis is made on advancing skills in prenatal ultrasound to pick anomalies/malformation early and thereafter the infant can be delivered in a tertiary center. At the same time emphasis is made to do CXR as soon as possible in any neonate presenting with progressive worsening of respiratory distress to diagnose on going disease early and proper management can be rendered. We also recommend CT/MRI scan whenever diaphragmatic hernia is suspected on CXR to rule out other congenital malformation that mimic CPAM.

Acknowledgements

We acknowledge both parents for agreeing to consent for academic purpose and publication

Authors' contributions
Rukhsar Osman: corresponding author, manuscript editing
Naomi Mwamanenge: literature search, manuscript preparation
Yaser Abdallah: manuscript review
Karim Manji: Manuscript review
Conflict of interest: None
Funding: None

References

1. Baral D, Adhikari B, Zaccarini D, Dongol RM, Sah B. Congenital pulmonary airway malformation in an adult male: a case report with literature review. Case Reports in Pulmonology. 2015;2015 (1):743452.
2. Sime H, Abera G, Mengistu A, Lamessa S. Congenital Pulmonary Airway Malformation (CPAM): A Case Report, Jimma University Medical Center, Southwest Ethiopia. Ethiopian Journal of Health Sciences. 2021;31(4).
3. Bolde S, Pudale S, Pandit G, Ruikar K, Ingle SB. Congenital pulmonary airway malformation: A report of two cases. World Journal of Clinical Cases: WJCC. 2015;3(5):470.
4. David M, Lamas-Pinheiro R, Henriques-Coelho T. Prenatal and postnatal management of congenital pulmonary airway malformation. Neonatology. 2016;110(2):101-15.
5. López-Díaz M, Antón-Pacheco JL, Gallego-Herrero C, Enguita -Vals A, Cano-Novillo I, Morante-Valverde R, et al. Diagnostic accuracy of imaging compared to histology in congenital lung malformations. Anales de Pediatría (English Edition). 2023;99(5):304-11.
6. Hsu JS, Zhang R, Yeung F, Tang CS, Wong JK, So M-T, et al. Cancer gene mutations in congenital pulmonary airway malformation patients. *ERJ Open Research. 2019;5(1).*
7. Ottomeyer M, Huddleston C, Berkovich RM, Brink DS, Koenig JM, Sobush KT. Early resection of a rare congenital pulmonary airway malformation causing severe progressive respiratory distress in a preterm neonate: a case report and review of the literature. BMC pediatrics. 2023;23 (1):238.
8. Wu ZJ, Narula N, Beneck D, Nguyen AB, Lee PC. Congenital pulmonary airway malformation. Clinical Pulmonary Medicine. 2016;23(5):227-30.
9. Hermelijn SM, Zwartjes RR, Tiddens HA, Cochius-den Otter SC, Reiss IK, Wijnen RM, et al. Associated anomalies in congenital lung abnormalities: a 20-year experience. *Neonatology. 2021;117(6):697 -703.*
10. Lim J, Han JH, Shin JE, Eun HS, Lee SM, Park MS, et al. Evaluation of Newborn infants with prenatally diagnosed congenital pulmonary airway malformation: a single-center experience. *Neonatal medicine. 2019;26(3):138-46.*
11. Mwamanenge NA, Mussa F, Nyamuryekung'e MK, Mkony M, Abdallah Y, Manji K. Late diagnosis of congenital diaphragmatic hernia: a case report. *Journal of Medical Case Reports. 2023;17(1):240.*
12. Hellmund A, Berg C, Geipel A, Bludau M, Heydweiller A, Bachour H, et al. Prenatal diagnosis and evaluation of sonographic predictors for intervention and adverse outcome in congenital pulmonary airway malformation. PloS one. 2016;11(3):e0150474.
13. Macchini F. Thoracoscopic resection of congenital pulmonary airway malformations: timing and technical aspects. *Journal of Thoracic Disease. 2020;12(8):3944.*
14. Farolfi A, Ghezzi M, Calcaterra V, Riccipetitoni G, Pelizzo G, Costanzo S, et al. Congenital lung malformations: clinical and functional respiratory outcomes after surgery. *Children. 2022;9 (12):1881.*
15. Peters NC, Hijkoop A, Hermelijn SM, van Schoonhoven MM, Eggink AJ, van Rosmalen J, et al. Prediction of postnatal outcome in fetuses with congenital lung malformation: 2-year follow-up study. *Ultrasound in Obstetrics & Gynecology. 2021;58 (3):428-38.*
16. Mehta PA, Sharma G. Congenital pulmonary airway malformation.