DE GRUYTER OPEN

J Biomed Clin Res Volume 8 Number 2, 2015

DOI: 10.1515/jbcr-2015-0168

Case Report

CONGENITAL CYSTIC ADENOMATOID MALFORMATION OF THE LUNG: A CASE REPORT AND REVIEW OF THE LITERATURE

Doroteya V. Malinova,
Penka L. Kolova,
Radoslav S. Radev[1]

Department of General and Clinical pathology,
Forensic Medicine and Deontology,
Medical Faculty,
Medical University, Varna
[1] Department of Surgical Diseases,
Medical Faculty, Medical University, Varna

Summary

Congenital cystic adenomatoid malformation (CCAM) is a rare abnormality of lung development. It was classified into 5 types by Stocker in 2002 and is also known under the name of congenital pulmonary airway malformation (CPAM). Cases are typically identified prenatally by routine ultrasonography screening. CCAM may present in the older child and adult as an incidental finding. The case presented is of a 21-year-old male patient who suffered from pulmonary infections with a recurrent productive cough 3-4 times the last 4 years. CT scanning of the thorax showed multiple cystic lesions in the right middle and lower lobes. The areas with the lesions were resected. The macroscopic and histological findings were typical for congenital cystic adenomatoid malformation type 2, which was the final diagnosis. Clinical presentations and prognosis depend on the type of lesion and its sequelae. The diagnosis is confirmed histologically.

Key words: congenital cystic adenomatoid malformation, types, prognosis

Corresponding Author:
Doroteya V. Malinova
Department of General and Clinical pathology,
Forensic Medicine and Deontology
Medical University Prof. Dr. Paraskev Stoyanov
55, Marin Drinov str.
Varna, 9002
e-mail: dmalinova@gmail.com

Received: November 01, 2014
Revision received: May 29, 2015
Accepted: December 01, 2015

Introduction

Congenital cystic adenomatoid malformation (CCAM) is a type of congenital malformation and refers to a group of malformations of the airways. There are different types of lesions (types 0-4), and some are associated with cystic areas and adenomatous overgrowth of the terminal bronchioles [1, 2]. Cases are typically identified prenatally by routine ultrasonography screening [3]. The lesion is usually unilateral (85%) with an equal incidence of microcystic or macrocystic disease, although bilateral cases have been reported [1-3]. CCAM may remain undiagnosed until discovered as an incidental finding later in life. However, its usual postnatal presentation is respiratory distress in the newborn period. This may be due to pulmonary hypoplasia, mediastinal shift, spontaneous pneumothorax, and pleural effusions secondary to hydrops. Recurrent chest infections may be a feature later in life [4]. Prenatal regression and complete prenatal resolution have also been described [5].

Epidemiology

Data from large population registries suggest an incidence of congenital lung cysts in the range of 1 per 8300 to 35 000 live births [6]. Large-cyst subtypes account for about 70% of congenital pulmonary airway malformations (CPAMs), or 2 to 8 per 100 000 live births.

Etiology and pathogenesis

The cause of CCAM is unknown. One theory holds that it is the result of bronchial structures failing to mature properly at the time the lungs begin to form - the fifth or sixth week of gestation. Another theory suggests that CCAM is due to an abnormal growth pattern of lung tissue that occurs as a result of bronchial obstruction [7]. Histologic studies reveal rapid vascular and epithelial growth within the tumor. Recent findings of accelerated cellular proliferation and decreased apoptosis within resected CCAM specimens further suggest a benign neoplastic development [1]. There is no known genetic cause for CCAM, and no cases of recurrence in a sibling or offspring have been reported. Increased cell proliferation and decreased apoptosis may cause malignant transformation. Pleuropulmonary blastoma, and bronchioloalveolar carcinoma in particular have been reported [7-11].

Case presentation

We present a case of a 21-year-old male patient who suffered from pulmonary infections with a recurrent productive cough 3-4 times the last 4 years. Computed tomography (CT) of the thorax showed multiple cystic lesions (Figure 1) in the right middle and lower lobes. The areas with the lesions were resected. Macroscopic and histological findings were typical for CCAM type 2, which was the final diagnosis. Gross inspection revealed multi-cystic lesion with partially solid and necrotic areas (Figure 2). Histology showed proliferation of bronchiole-like structures and macro- and microcysts lined by columnar or cuboidal epithelium and absence of cartilage and bronchial glands. Squamous metaplasia and inflammatory cells were observed in some areas (Figure 3, 4).

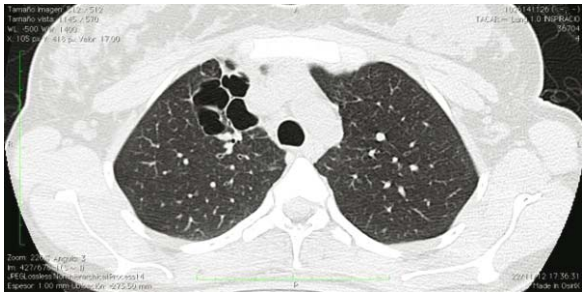

Figure 1. CT of the thorax of a patient with CCAM type 2 – multiple cystic lesions of less than 1 cm in diameter in the right middle and lower lobes of the lung

Figure 2. Gross inspection – multi-cystic lesion with partially solid and necrotic areas (cysts are less than 1 cm in diameter)

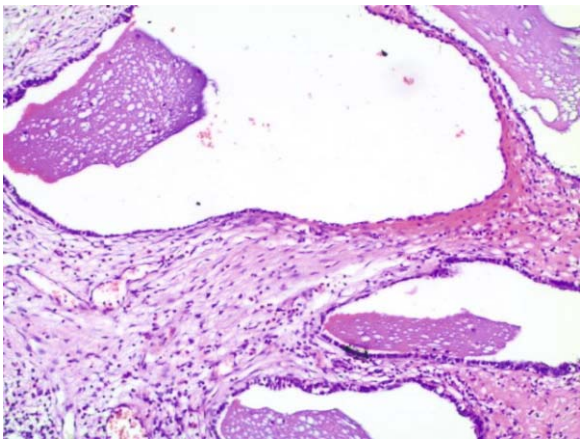

Figure 3. Macro- and microcysts lined by columnar or cuboidal epithelium. Walls of the cysts are composed of connective tissue and smooth muscle cells. There are no communications between the cystic structures (Hematoxylin and eosin stain, 20x10)

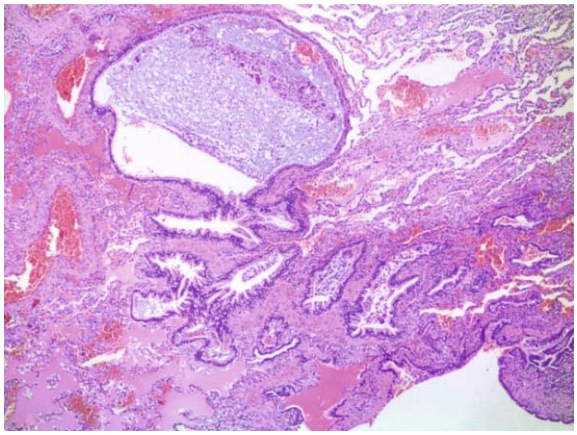

Figure 4. Bronchiole-like structures lined by columnar or cuboidal epithelium with walls composed of connective tissue and smooth muscle cells, without cartilage and glands. In the lumen of the structures there are inflammatory cells. In the surrounding area hyperemia and inflammatory cells (Hematoxylin and eosin stain, 20x10)

Discussion

Stocker [7, 12, 13] classified these congenital lung lesions based on the site of their origin and labelled them from 0 to 4 to indicate their progression down the airways. Type 0, formerly called acinar dysplasia [14], is reffered to as bronchial and is associated with other malformations. Type I is characterised by large cysts - up to 10 cm in diameter, and is described as bronchial/bronchiolar type. It constitutes about 50% of postnatal cases of CCAM. Type I cysts are lined by pseudostratified cilliated cells. The prognosis is usually good. Type II lesions are called bronchiolar and comprise 40% of postnatal cases. Type II cysts are small (about 1 cm in diameter) and numerous. They resemble dilated bronchioles and are separated by normal alveoli. These cysts are lined by cuboidal, ciliated, or columnar epithelial cells. Type II lesions are often associated with other congenital malformations and the outcome depends on the severity of these anomalies. Common malformations associated with type II include renal agenesis and dysgenesis, cardiac anomalies – tetralogy of Fallot, persistant truncus arteriosus, diaphragmatic hernia, small intestine anomalies - jejunal atresia, skeletal anomalies [8, 14]. Type III lesions are called bronchiolar/alveolar duct and they comprise 10% of the cases. Grossly, this type is characterised by large microcystic-appearing masses that may cause mediastinal shifting. Histologic findings include bronchiolar structures separated by alveoli. Bronchiloles are lined by ciliated cuboidal epithelium and alveolar spaces between them – by nonciliated cuboidal epithelium. The outcome of type III lesions is variable[15]. Type IV cysts are described as peripheral. They account for 15% of postnatal cases. Type IV cysts are large (up to 10 cm in diameter) and lined by flattened epithelial cells. Histologically they may resemble cystic pleuropulmonary blastoma [11, 13, 16]. Stocker (1994) suggested that the term congenital pulmonary airway malformation was more accurate, because of the large variety of anomalies included in this classification. However, CCAM and CPAM are both used today [13].

In infants and children, malformations communicate with the tracheobronchial tree, creating a valvular mechanism, which causes hyperinflation of the cystic zone during inspiration and less deflation during expiration. Enlarged cysts compressing the normal pulmonary structure lead to the respiratory symptoms. Glandular tissue proliferation may cause mucin accumulation and recurrent infections. The majority of clinical presentations of CPAM atdifferent ages have been reported as respiratory distress and failure in infants, and repeated airway infections in children [17-19]. In adults, it is also most commonly presented with repeated airway infections. To the best of our knowledge, there have been fewer than 60 reports of CPAM in patients aged 18 and over. The association between CPAM and malignancy has also been well documented. Malformation and proliferation cause hamartomas over the tracheobronchial tree. Type 1 CPAM may involve malignant transformation of mucinous bronchioloalveolar carcinoma [10, 20], and type 4 CPAM requires examination of the entire lesion to exclude pleuropulmonary blastoma [7, 21]. Even though malignant transformation is rare, the prognosis is very different from that in benign cystic lesions. Surgical resection is the gold standard of management for CPAM for both pathological diagnosis and treatment.

In conclusion, CPAM can present at any age and the clinical appearances vary from immediate postnatal respiratory failure to an accidental finding on chest radiography. Malignancy and frequent airway infections are major concerns in these patients. Surgical excision is recommended to make a definite diagnosis and rule out hidden malignancies. It is also the treatment of choice.

References

1. Wilson RD, Hedrick HL, Liechty KW, Flake AW, Johnson MP, Bebbington M, et al. Cystic adenomatoid malformation of the lung: review of genetics, prenatal diagnosis, and in utero treatment. Am J Med Genet A. 2006;140(2):151-5.
2. Kotecha S, Barbato A, Bush A, Claus F, Davenport M, Delacourt C, et al. Antenatal and postnatal management of congenital cystic adenomatoid malformation. Paediatr Respir Rev. 2012;13(3):162-70; quiz 170-1.
3. Laberge JM, Flageole H, Pugash D, Khalife S, Blair G, Filiatrault D, et al. Outcome of the prenatally diagnosed congenital cystic adenomatoid lung malformation: a Canadian experience. Fetal Diagn Ther. 2001;16(3):178-86.
4. Luján M, Bosque M, Mirapeix RM, Marco MT, Asensio O, Domingo C. Late-onset congenital cystic adenomatoid malformation of the lung. Embryology, clinical symptomatology, diagnostic procedures, therapeutic approach and clinical follow-up. Respiration. 2002;69(2):148-54.
5. Sauvat F, Michel JL, Benachi A, Emond S, Revillon Y. Management of asymptomatic neonatal cystic adenomatoid malformations. J Pediatr Surg. 2003;38(4):548-52.
6. Shanti CM, Klein MD. Cystic lung disease. Semin Pediatr Surg. 2008; 17:2-8.
7. Stocker JT. Cystic lung disease in infants and children. Fetal Pediatr Pathol. 2009;28:155-84.
8. Bromley B, Parad R, Estroff JA, Benacerraf BR. Fetal lung masses: Prenatal course and outcome. J Ultrasound Med. 1995;14: 27-936.
9. Di Prima FA, Bellia A, Inclimona G, Grasso F, Teresa M, Cassaro MN. Antenatally diagnosed congenital cystic adenomatoid malformations (CCAM): Research Review. J Prenat Med. 2012;6(2):22-30.
10. Ramos SG, Barbosa GH, Tavora FR, Jeudy J, Torres LA, Tone LG, et al. Bronchioloalveolar carcinoma arising in a congenital pulmonary airway malformation in a child: case report with an update of this association. J Pediatr Surg. 2007; 42:E1-4.
11. MacSweeney F, Papagiannopoulos K, Goldstraw P, Sheppard MN, Corrin B, Nicholson AG. An assessment of the expanded classification of congenital cystic adenomatoid malformations and their relationship to malignant transformation. Am J Surg Pathol. 2003;27:1139-46.
12. Stocker JT, Madewell JE, Dark RM. Congenital cystic adenomatoid malformation of the lung: Classification and morphologic spectrum. Hum Pathol. 1977;8: 155-71.
13. Stocker JT. Congenital and developmental diseases. In: Dail DH, Hammer SP, editors. Pulmonary pathology. 2nd ed. New York: Springer; 1994. p 174-80.
14. Davidson LA, Batman P, Fagan DG. Congenital acinar dysplasia: a rare cause of pulmonary hypoplasia. Histopathology. 1998;3:57-59.
15. Adzick NS, Harrison MR, Glick PL, Golbus MS, Anderson RL, Mahony BS, et al. Fetal cystic adenomatoid malformation: Prenatal diagnosis and natural history. J Pediatr Surg. 1985;20:483-8.
16. Thorpe-Beeston JG, Nicolaides KH. Cystic adenomatoid malformation of the lung: prenatal diagnosis and outcome. Prenat Diagn. 1994;14(8): 677-88.
17. Adzick NS. Management of fetal lung lesions. Clin Perinatol. 2003;30:481-92.
18. Huang HJ, Talbot AR, Liu KC, Chen CP, Fang HY. Infected cystic adenomatoid malformation in an adult. Ann Thorac Surg. 2004; 78:337-9.
19. Oh BJ, Lee JS, Kim JS, Lim CM, Koh Y. Congenital cystic adenomatoid malformation of the lung in adults: clinical and CT evaluation of seven patients. Respirology. 2006;11:496-501.
20. West D, Nicholson AG, Colquhoun I, Pollock J. Bronchioloalveolar carcinoma in congenital cystic adenomatoid malformation of lung. Ann Thorac Surg. 2007;83:687-9.
21. Oliveira C, Himidan S, Pastor AC, Nasr AD, Taylor G, Yanchar NL, et al. Discriminating preoperative features of pleuropulmonary blastomas (PPB) from congenital cystic adenomatoid malformations (CCAM): a retrospective, age-matched study. Ann Thorac Surg. 2011;21:2-7.