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Caso clínico

Cystic adenomatoid malformation in a 2-month-old Mexican infant. Case report and review of the literature

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RESUMEN

Antecedentes: las malformaciones adenomatoideas quísticas congénitas son anomalías hamartomatosas en el tejido pulmonar que abarcan la vía aérea distal. Representan 25% de las malformaciones pulmonares congénitas.
Reporte del caso: se comunica el caso de una paciente de dos meses de edad referida por diagnóstico presunto de infección en vía aérea baja y exposición a tuberculosis. Nació por vía cesárea sin complicaciones y con peso y talla adecuados a su edad gestacional; lactó y fue complementada con fórmula. Su desarrollo psicomotriz fue normal, tenía sus vacunas al corriente y su madre tuvo una enfermedad del colágeno indeterminada. El examen físico y los estudios de laboratorio fueron normales. Los estudios radio y tomográficos revelaron una masa gruesa moderadamente radiopaca en la pared torácica con bordes bien definidos localizada en el lóbulo inferior derecho con tres círculos radiolúcidos en su interior. Los espacios intercostales estaban crecidos, y el corazón y el mediastino desplazados a la derecha. Se le practicó lobectomía superior derecha. El estudio anatomopatológico reveló malformación adenomatoidea quística congénita tipo I.
Conclusión: la evaluación prenatal y el diagnóstico apropiado son vitales para un resultado positivo. Aun cuando es una enfermedad rara, es importante que el médico evalúe su diagnóstico durante el ultrasonido de las pacientes embarazadas. La paciente se recuperó sin complicaciones.
Palabras clave: malformación adenomatoidea quística congénita, masa pulmonar, malformación pulmonar congénita.

ABSTRACT

Background: Congenital cystic adenomatoid malformation (CCAM) is a multicystic hamartomatous abnormality of lung tissue that involves the distal airway. It represents 25% of all congenital lung malformations.
Case report: We describe the case of a 2 month old female referred with a presumptive diagnosis of lower respiratory tract infection and positive tuberculosis exposure. The patient was delivered by cesarean section after an uneventful pregnancy with adequate weight and height for gestational age. The child was breast fed with formula supplementation; psychomotor development was unremarkable. Her immunizations were up to date; her mother has an unspecified collagen disease. Physical examination and laboratory tests were unremarkable. Chest X-ray and CT scan revealed a moderately radiopaque thick walled mass with well defined borders located in the right lower lobe with three radiolucid circles in its interior. The intercostal spaces were enlarged, the heart and mediastinum displaced to the right. The patient underwent a right upper lobectomy. Pathology reported a type I CCAM.
Conclusion: Prenatal screening and proper diagnosis are vital for a positive outcome. Although a rare pathology, it is important for physicians to consider the diagnosis each time they perform an ultrasound on a pregnant patient. Our patient's recovery was uneventful.
Key Words: Congenital cystic adenomatoid malformation, lung mass, congenital lung malformation.

Congenital Cystic Adenomatoid Malformation (CCAM) is a multicystic hamartomatous abnormality of lung tissue with proliferation of bronchial structures that involves the distal airway.[1-3] It represents 25% of all congenital lung malformations.[4] The reported incidence is 1:25,000 to 1:35,000 pregnancies.[1] Ch'in and Tang first described this pathology in 1949,[5] but it was not until 1977 that Stocker et al developed a classification based on clinical, pathological and radiological features (Table 1).[6,7] CCAM occurs sporadically, with no association to age or ethnicity. Recent research suggests that alcohol consumption during the 8th to 10th week of gestation may cause CCAM by inducing cytochrome P450 which consequently decreases the production of retinoic acid. This causes a decrease in lung mesenchyme during the early stages of lung development. Hoxb-5 gene and protein expression has also been implicated.[3] We report the case of a 2 month old female, who was referred with a presumptive

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Table 1. Modified Stocker CCAM classification[6,15]

Type	*Frequency*	*No. of lesions*	*Size*	*Epythelium*	*Outcome*
I	50%	Single or multiple (<4)	2-10 cm	Pseudostratified ciliated	Good
II	40%	Multiple	< 2 cm	Ciliated or columnar	
III	10%	Multiple cystic (or solid)	microscopic	Non-ciliated cuboidal	Worse

diagnosis of lower respiratory tract infection and positive tuberculosis exposure.

CASE REPORT

The patient is the 2 month old female child of a 29 year old woman (G-3, P-0,C-1, A-1) with a history of an unspecified collagen disease of two years evolution with positive antiphospholipid antibodies treated with heparin. The child was born full term by cesarean section after an uneventful pregnancy with adequate weight and height for gestational age. She had been breast fed with formula supplementation and had normal psychomotor development. Her immunizations were up to date.

The patient was seen by her pediatrician for a productive cough of 15 days evolution, initially diagnosed and treated as bronchiolitis. The patient's brother had developed cough five days previous to this event that resolved with supportive treatment. The family also recently discovered that the housekeeper had a history of pulmonary tuberculosis. The patient's condition worsened during the next three days. The cough intensified leading to emesis. Fever, trouble feeding and malaise were denied. She was admitted to the hospital for further evaluation.

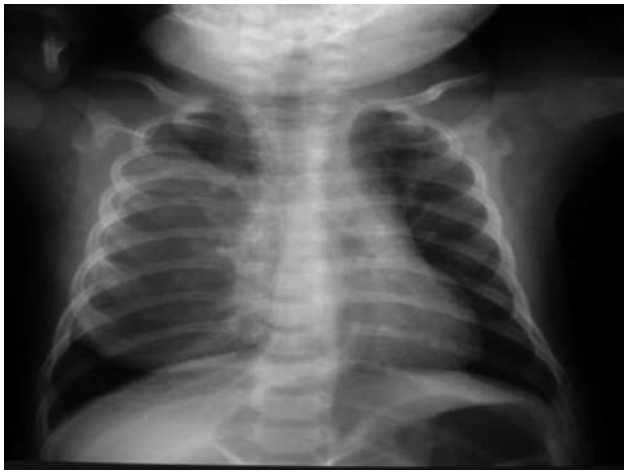

Figure 1. Chest X-Ray, front view. Radiopaque mass located in the right lung with well-defined borders. Intercostal spaces in the right hemithorax are increased.

On physical examination her vital signs were HR 155 / min, RR 44 / min, temperature 37.5° C, oxygen saturation was 95% breathing room air; weight was 4, 430 grams. She was well hydrated with generalized pallor of the skin and mucosa, and hyaline rhinorrea. No enlarged lymph nodes were found. Chest examination revealed rhythmic heart sounds with good intensity and no evidence of murmurs. Peripheral pulses were present and normal. Bilateral soft crackles with decreased bronchovesicular sounds were detected. There was no evidence of respiratory distress. Complete blood count revealed a red blood cell count of 2.73 M / mL; a hemoglobin of 8.10 g / dL, and a hematocrit of 22.5%; a leukocyte count of 12.10 K / mL; a platelet count of 920 K / mL. Other laboratory parameters were within normal range. Chest x-ray and a CT scan revealed a moderately radiopaque mass located in the right lower lobe that elevated and forwardly displaced the minor fissure occupying 60% of the right lung and increasing the intercostal spaces, pushing the heart and mediastinum to the right. The borders were well defined. The anterior border had a thick wall. Three radiolucid circles were found in its interior (Figures 1 and 2) The patient was programmed for surgery during which a right upper lobectomy and resection of the mass by a right posterolateral thoracotomy at the fourth intercostal space was performed. The postoperative course was uneventful with a rapid recovery. The mass was sent to pathology that reported a Type I Congenital Cystic Adenomatoid Malformation.

DISCUSSION

Although its exact etiology is uncertain,[8] there are several theories that explain the origin of CCAM.

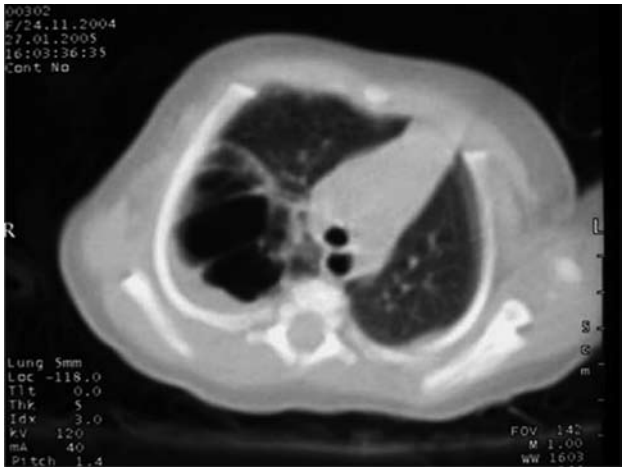

Figure 2. Chest CT-scan. Right lung mass composed by various cysts of different sizes. There is left mediastinal deviation.

One of the most widely accepted states that CCAM is caused by a failure of maturation of bronchiolar structures and concomitant overgrowth of mesenchymal elements.[9-11] The time frame for its formation is between the 5th and 7th week of gestation,[9,11-14] during the pseudoglandular stage of lung development.[9,12] CCAM is slightly more common in males (1.8:1),[9] but there is no direct association for ethnicity or geography.[12,15] CCAM can be seen in any part of the lung, even in a bilateral presentation, but several reviews concur that the most common presentation (85-90%) is unilateral, with a predilection for the basal lobes as a single lesion;[9] another review concludes that both sides are equally affected.[13] In a series of 15 cases 10 were located on the right side.[11]

The clinical presentation varies depending on the infant's age[16] and the size of the lesion.[11] Patients can be asymptomatic[11] or start with a variety of signs and symptoms including respiratory distress, cough, with or without cyanosis, stridor, recurrent pulmonary infections, hemoptysis, dysphagia, spontaneous pneumothorax, and life-threatening events of respiratory failure.[14,16,17] Respiratory distress is more frequent in patients < 1 year of age and recurrent pulmonary infections are more frequent in patients > 1 year of age.[11]

It is important to recognize that CCAM, especially type 2, may be accompanied by other malformations such as cardiac malformation, renal agenesis or dysgenesis, jejunal atresia, and diaphragmatic hernia.[9] The symptoms caused by the tumor, as well as complications that may arise, are secondary to compression of intrathoracic structures. The three main complications associated with CCAM are polyhydramnios (35%), hydrops fetalis (85%), and mediastinal shift.[1,11,13,14] Polyhydramnios is caused by the overproduction of amniotic fluid by tumor tissue and decreased ability to swallow due to compression of the esophagus. Hydrops fetalis is caused by compression of the superior vena cava and heart failure caused by mediastinal shift.[13] The use of prenatal ultrasound (US) has increased the detection of asymptomatic cases of CCAM.[18] Therefore, it is reasonable to use it as an initial screening tool to study any thoracic abnormality. It is also helpful to detect the size, type and location of the lesion as well as for searching for signs of complications.[19,20] US has an accuracy of 72% in identifying and correctly diagnosing a fetal chest lesion. It can correctly diagnose CCAM, based on the Stocker classification in 57% of cases, incorrectly in 29% and fail in 14%.[11] The mean gestational age at diagnosis by ultrasound has been reported as 22-23 weeks (range: 16-40).[1,13] Approximately 85% of the cases are diagnosed before 25 weeks of gestation. Spontaneous tumor regression has been reported in several reviews, ranging from 6% to 56%.[1,13,18,19,21]

Another helpful tool is a chest CT scan which shows the extent of the lesion, depicts multicystic areas, identifies air fluid levels, and demonstrates unsuspected involvement of surrounding tissues. It is also useful for surgery planning.[21] A CT scan of the chest, besides its ability to correlate with the histological classification, has the advantage of detecting lesions that do not appear sonographically or radiologically, as well as differentiating CCAM from other lesions.[11] Despite the technological advances of imaging studies, definitive diagnosis cannot be made with certainty.[1,17,21] Those lesions that escaped identification by prenatal screening present clinically in the first 2 years of life.[11]

Early diagnosis and prompt surgical treatment remain the approaches of choice to achieve optimum outcome.[14] Selection of a specific therapeutic course of action will depend on the presence or not of any complications during pregnancy, type of lesion and gestational age.[19]

Based on the literature review we developed an algorithm for the diagnosis and treatment of CCAM (Figure 3).[19-21] Lobectomy remains the gold standard of treatment because it has proven to prevent residual disease, recurrence[12,18] and malignant transformation.[18,22] Differential diagnosis includes congenital diaphragmatic hernia, bronchogenic cyst, pulmonary sequestration, bronchial atresia.[9]

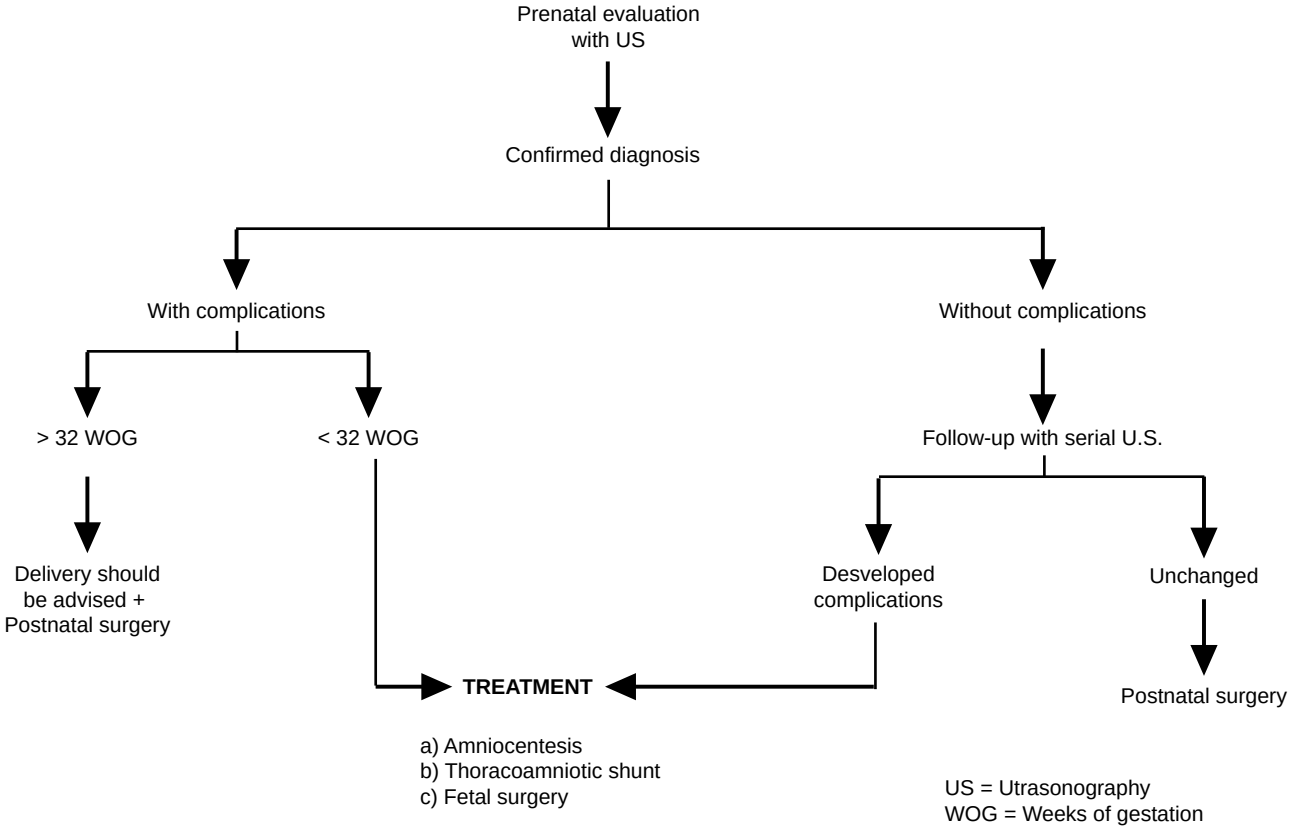

Figure 3. CCAM diagnosis and treatment flowchart, based on literature review.[19,20,21]

Prognosis is variable, from perinatal death to spontaneous in uterus regression with no neonatal morbidity,[13] but it depends primarily on in uterus evolution and the presence of other malformations.[1] Gestational age at the moment of diagnosis does not impact overall outcome.[1] Prenatal and postnatal large lesions or mediastinal shift do not necessarily mean a poor prognosis, but they do correlate with clinical symptoms.[11] The life expectancy after thoracocentesis or thoracoamniotic shunt is 68%,[19] compared to 100% survival for uncomplicated CCAM cases treated with postnatal surgery.[13]

CONCLUSION

Prenatal US screening and correct diagnosis is vital for the outcome of the CCAM patient. Although it is a rare pathology, it is important for physicians to keep it in mind each time they perform an ultrasound on a pregnant woman. Our patient, as well as other cases reported, had an uneventful recovery and was discharged 10 days after surgery with no further complications.

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