



## Congenital cystic adenomatoid malformation with prenatal diagnosis: case report

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### Summary

Congenital cystic adenomatoid malformation is a hamartomatous malformation accepted as an embryonic differentiation anomaly characterized by replacement of normal lung parenchyma by cysts of various sizes and numbers. The primigravid presented on her 24<sup>th</sup> week with the ultrasound findings of a fetus with unilateral cysts in left lung. Thoracoamniotic shunting was inserted at a mean gestational age of 24. Patient was born due to premature rupture of membranes and preterm delivery at a mean gestational age of 25. We present a case of congenital cystic adenomatoid malformation type I in a newborn who died in twelve hours after birth and discuss about pre- and postnatal diagnostic and therapeutic possibilities. (*Turk Arch Ped* 2012; 47: 290-293)

**Key words:** Congenital cystic adenomatoid malformation, newborn

### Introduction

Congenital cystic diseases of the lung and mediastinum include congenital cystic adenomatoid malformation (CCAM), bronchopulmonary "sequestration", congenital lobar emphysema, bronchogenic cyst, esophageal duplication cysts and neurogenic cysts (1).

Congenital cystic adenomatoid malformation is an embryonic developmental disorder characterized by cystic enlargement and overgrowth of terminal bronchioles which are surrounded by various epithelial cells. It occurs at the 6-8<sup>th</sup> gestational week (GW). It is observed with an equal frequency in both lungs and its incidence ranges between 1/10 000 and 1/35 000. The lesions usually tend to involve a single lobe and especially the lower lobe. The incidence of the disease does not vary according to race or gender (2).

Congenital cystic adenomatoid malformation is a rare but life-threatening lung anomaly. Cysts which grow and enlarge can compress the lungs, large vessels and esophagus. As a result of this it can lead to mediastinal shift, hydrops and polyhydramnios in fetal life and life-threatening respiratory distress in the postnatal

period. However, most patients have no complaints at birth and lung graphies taken in the postnatal period are normal (3).

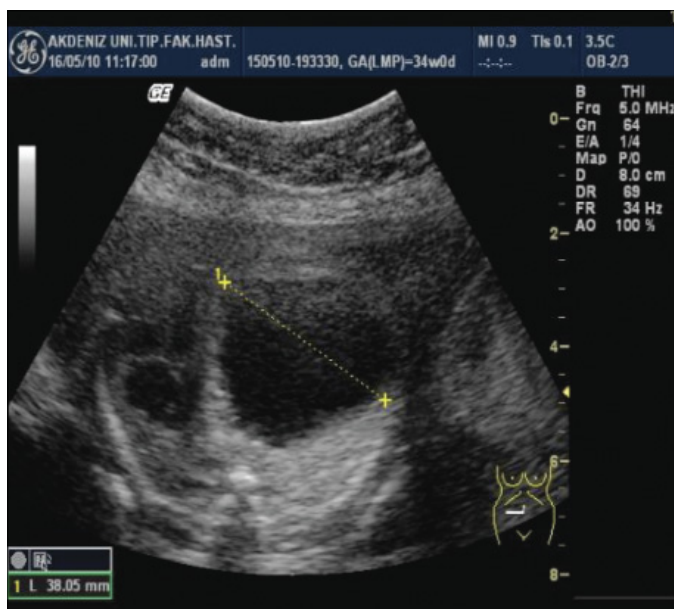
We would like to present a patient with CCAM who was diagnosed in the intrauterine period and underwent thoracoamniotic shunting and who was lost in the first day of life with severe respiratory distress.

### Case

Our patient was born at the 25<sup>th</sup> gestational week according to the last menstruation date by cesarean section with a birth weight of 670 g from the first pregnancy of a 20-year-old mother. He was followed up with the diagnoses of polyhydramnios and hydrops fetalis in the prenatal period. On ultrasonographic examination (USG) performed at the 24<sup>th</sup> gestational week, a uniloculated and pure cystic lesion with regular contours and a relatively thick wall including air-fluid level which caused shifting of the middle line and mediastinal structures towards the right side was observed in the left hemithorax. The largest size of the cyst was measured to be 4x3,5x3,8 cm (Picture 1). Hydrops fetalis due to type 1 CCAM was found on ultrasonography. Thoracoamniotic

shunt was inserted at the 24<sup>th</sup> gestational week by the department of obstetrics and gynecology to ameliorate mass compression. The patient was born due to premature rupture of membranes and preterm delivery by urgent cesarean section one week after the procedure. During cesarean section, it was observed that the place of the thoracoamniotic shunt catheter was not good and the catheter floated in the amniotic fluid.

The patient who did not cry immediately at birth and who had APGAR scores of 3 and 5 at the first and 5<sup>th</sup> minutes was intubated and internalized in the intensive care unit. On lung graphy, a cystic lesion which shifted the mediastinum towards the right side was observed in the left hemithorax and atelectasia was present in the right lung because of compression (Picture 2). On physical examination, the body weight was found to be 670 g (3-10<sup>th</sup> percentile), the height was found to be 32 cm (3<sup>rd</sup> percentile), the head circumference was found to be 26 cm (3<sup>rd</sup> percentile), the respiratory rate was found to be 66/min and the oxygen saturation was found to be 78%. No additional pathologic physical examination finding was observed except for premature delivery. Laboratory tests revealed that complete blood count, electrolytes, renal and hepatic function tests and CRP level were normal. The baseline blood gases were as follows: pH: 7.17, pCO<sub>2</sub> 78.6 mmHg, pO<sub>2</sub> 54 mmHg and HCO<sub>3</sub> 15.1 mmol/L, BE:-7 mmol/L. On thoracic computed tomography (CT), a cystic lesion with a relatively thick wall including microcysts in the lower part and air-fluid level was observed in the left hemithorax causing shifting of the middle line and mediastinal structures towards the right side. The largest transaxial size of the cyst was measured to be 4.5x3.5x5 cm (Picture 3). Thoracosynthesis was performed in the left hemithorax of the patient whose oxygen saturation could not



Picture 1. A large cystic mass (4x3.5x3.8 cm) leading to shifting of the heart and mediastinum towards the right side is observed in the left thorax on USG performed at the 24<sup>th</sup> gestational week

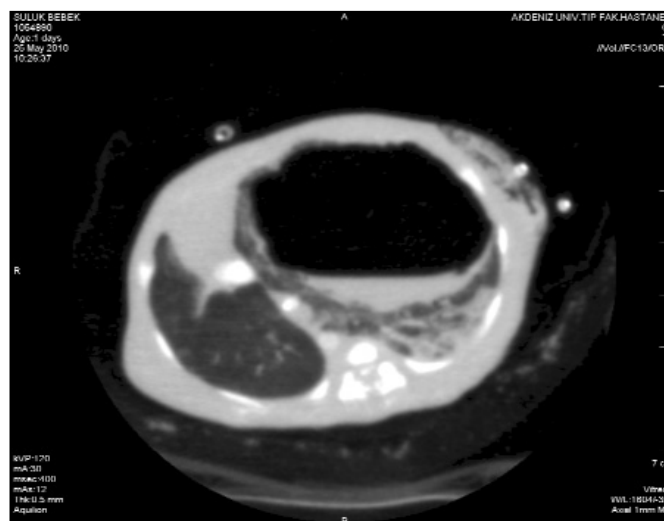
be increased; 5 cc serous fluid and 20 cc air was derived and a thoracic catheter was placed. No change in the cyst's diameter was observed after placing the chest tube. Surgical intervention was planned, but the patient was lost in the first 12 hours because of respiratory distress.

## Discussion

Congenital cystic malformation was classified in three types by Stocker et al. (4) in 1977 based on clinical, macroscopic and microscopic criteria. Type I is the most common type (50-60%) and is composed of one or multiple cysts with a diameter of 2-10



Picture 2. Radiologic appearance of the cystic lesion which leads to shifting of the mediastinum towards the right side and includes the whole left lung on PA chest graphy



Picture 3. Cystic lesion in the left hemithorax with the largest size measured to be 4.5x3.5x5 cm which leads to shifting of the middle line and mediastinal structures towards the right side on thoracic CT

cm. Its wall is layered by multiple pseudo-stratified cylindrical epithelium. Type II occurs with a rate of 30-40% and includes multiple small cysts layered by cuboid cylindrical epithelium with a diameter of 0.5-2 cm. Type III occurs with a rate of 10% and has the worst prognosis.

Sonography is the primary method in evaluation of fetal thorax. The classification made by Stocker et al. (4) according to anatomic and clinical variations can be used as sonographic typing. In our patient, the diagnosis was made by USG findings in the prenatal period and was found to be compatible with type I CCAM (5).

With use of prenatal imaging methods CCAM is diagnosed more frequently. When a mass is found in the lung in fetal life, its localization, size, dimensions and appearance should be carefully examined. The origin of the vascular structure of the mass should be determined by Doppler USG to differentiate it from pulmonary sequestration. Patients who are diagnosed as CCAM by USG in the prenatal period should be followed up closely in terms of development of fetal hydropic changes which indicate fetal mortality. In these patients, problems including polyhydramnios, lung hypoplasia and mediastinal shift can be determined by USG (2). Hydrops fetalis, lung hypoplasia due to cystic compression, polyhydramnios and mediastinal shift were found in our patient.

Development of hydrops due to congenital cystic adenomatoid malformation can be predicted in the prenatal period by calculating CCAM volume ratio (CVR) on USG. Cystic volume is found by multiplying the length, width and depth of the cyst measured on USG by 0.523 (length x width x depth x 0.523). CCAM volume ratio reflects the ratio of the cystic volume to head circumference. When CCAM volume ratio was <1.6, the survival rate and the rate of development of hydrops were found to be 94% and 3%, respectively (6). In our patient, CCAM volume ratio was found to be 1.78 and hydrops fetalis was present. Therefore, CCAM volume ratio appears to be useful in determining the risk of development of hydrops.

In approximately 15% of the patients with congenital adenomatoid malformation, the diameter of the cyst decreases as the gestational age increases and the reason for this is unknown. The cyst reaches its largest size at the 28<sup>th</sup> gestational week and decreases in size in approximately 20% of the patients after the 29<sup>th</sup> gestational week (3). Since our patient was born at the 25<sup>th</sup> gestational week, the prognosis of the cyst could not be followed up.

Mortality depends on the size of the primary lesion. In a study performed in Canada in 48 patients with CCAM, the mortality rate was reported to be 10% (7). 10 of these patients were reported to be lost after birth, 8 patients were reported as spontaneous or voluntary abortion and no mortality was reported in 30 patients. It has been observed that large lesions have a higher risk to develop mediastinal shift, vascular failure, polyhydramnios, hydrops fetalis and lung hypoplasia due to the effect of compression. Type III lesions have a poorer prognosis because of a tendency to become larger. Other factors which affect the prognosis include bilateral lung involvement, premature delivery and accompanying

severe malformations. Anomalies are usually associated with type II CCAM. Generally, renal, cardiac, intestinal and skeletal anomalies accompany (7).

The follow-up and diagnosis of fetal CCAM depends on the time of diagnosis, the size of the cyst and presence of findings related to the compression of the cyst. In fetuses in whom CCAM alone is present without fetal hydrops, follow-up with serial USG and term delivery are recommended. If sole CCAM and hydrops fetalis are present, extrauterine "intrapartum" surgical treatment is performed after the 32<sup>nd</sup> gestational week and thoracoamniotic shunt or open fetal surgery are recommended before the 32<sup>nd</sup> gestational week. Thoracoamniotic shunt may not be efficient in CCAM with a predominantly solid lesion and open fetal surgery is recommended in this case (8). In our patient, thoracoamniotic shunt was inserted, since the gestational age was younger than the 32<sup>nd</sup> gestational week, but the shunt was not functional, since the catheter was displaced.

The general rule in the postnatal therapeutic approach in CCAM is as follows: it is reported that surgical procedure may not be needed in patients without complaints. In patients with complaints, surgical treatment is controversial and close follow-up of these patients is recommended. Çetinkaya et al. (9) reported that the cysts completely disappeared at the 5<sup>th</sup> month after birth with postnatal traditional treatment in a patient with type I CCAM and emphasized the importance of traditional treatment. Özdemir et al. (10) emphasized the importance of early surgical treatment in terms of prognosis in a patient with CCAM who was operated on the 12<sup>th</sup> day because of symptoms (10). Even if CCAM is asymptomatic in the postnatal period, it is recommended that the masses should be excised completely because of the risk of recurrent infection and development of carcinoma (8).

Our patient who was evaluated as type I CCAM and was expected to have a good prognosis was lost in the first 12 hours. Conclusively, the type of CCAM alone does not indicate prognosis. Secondary pathologies (hydrops fetalis, polyhydramnios) and CVR should be carefully evaluated in association with the type of CCAM.

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