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Case Report

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Bilateral type 1 congenital cystic adenomatoid malformation: a case report

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Abstract

Objective: It is aimed to present a case of bilateral type 1 congenital cystic adenomatoid malformation which is prenatally diagnosed, and to discuss the management in these cases.

Case: A 27-year-old, gravida 4, parity 2, abort 1 patient was referred to our clinic with an initial diagnosis of hydrops fetalis at 25+5 weeks gestation of pregnancy. In the evaluation of fetus, it was detected by ultrasonography that bilateral pulmonary multilocular anechoic cystic lesions which were measured as maximum 24x26 mm were present, cardiac axis shifted to left, and common ascites were present. No additional anomaly was detected during the examinations of other systems. After the diagnosis of type 1 congenital cystic adenomatoid malformation, parents were informed about the fetal prognosis and termination of pregnancy was put forward as an option. Diagnosis was confirmed by the postmortem pathological examination.

Conclusion: The differential diagnosis of type 1 congenital cystic adenomatoid malformation and the presence of concomitant anomalies are important for the management of these cases. The prognosis is poor in cases with bilateral type 1 congenital cystic adenomatoid malformation accompanied by early hydrops fetalis, and the termination of pregnancy can be offered.

Key words: Bilateral congenital cystic adenomatoid malformation, prenatal diagnosis, management.

Bilateral Tip 1 konjenital kistik adenomatoid malformasyon: Olgu sunumu

Amaç: Prenatal tanı almış bilateral Tip 1 konjenital kistik adenomatoid malformasyon olgusunun sunulması ve yönetiminin tartışılması amaçlanmıştır.

Olgu: Yirmi yedi yaşında gravida 4, parite 2, abort 1 olan gebe 25+5 gebelik haftasında hidrops fetalis ön tanısı nedeniyle kliniğimize refere edilmiştir. Fetüsün değerlendirilmesinde bilateral akciğerlerde en büyüğü 26x24 mm boyutlarında multiloküler anekoik kistik yapılar, kalp aksının sola kaydığı ve yoğun assit saptandı. Diğer sistemlerin muayenesinde ek anomali saptanmadı. Tip 1 konjenital kistik adenomatoid malformasyon ön tanısı konularak; aile fetüsün prognozu açısından bilgilendirildi ve gebeliğin terminasyonu bir seçenek olarak sunuldu. Postmortem patoloji incelemesinde de tanı doğrulandı.

Sonuç: Tip 1 konjenital kistik adenomatoid malformasyon olgularının ayırıcı tanısı ve eşlik eden diğer anomalilerin varlığı yönetim açısından önemlidir. Erken fetal hidropsun eşlik ettiği bilateral Tip 1 konjenital kistik adenomatoid malformasyon olgularında prognoz kötü olup; gebeliğin tahliyesi önerilebilir.

Anahtar sözcükler: Bilateral konjenital kistik adenomatoid malformasyon, prenatal tanı, yönetim.

Introduction

Congenital cystic adenomatoid malformation (CCAM) is a hamartomatous lung lesion characterized by the proliferation of terminal bronchioles and abnormal alveolar development.[1,2] Its etiology has not been revealed yet, and it has been found out that it has a role in the pathology of increased apoptosis and that it is associated with HOXB5, FGF7, and PDGFB genes.[3,4] The malformation was first grouped in three types according to the sizes of cysts by Stocker in 1977.[5]

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In 2002, Stocker updated CCAM classification as five types by adding Type 0 and Type 4. Type 0 is small cystic lethal lesions arising from trachea and bronchus, and it is quite rare. Type 1 consists of 50-70% of CCAM cases and it includes cystic lesions which are 3-10 cm in size arising from distal bronchus and proximal bronchioles. Type 2 is the group having 0.5-2 cm lesions arising from terminal bronchioles and/or accompanied by solid lesions. Type 2 is 15-30% of CCAM cases and it is the group which is the most associated one (60%) with other system anomalies. Type 3 is caused by acinar tissue and includes microcystic solid hyperechogenic lesions; it constitutes 5-10% of CCAM cases. Type 4 is caused by alveolar tissue and it includes ≥10 cm cystic lesions; it is 5-15% of CCAM cases and it is particularly associated with pleuropulmonary blastoma.[6]

Adzick et al. categorized CCAM into two groups according to their ultrasonographic views, which are microcystic (<5 mm cystic or solid lesions) and macrocystic (>5 mm cystic lesions) types.[7] Most of the CCAM cases are unilateral, and they are lesions limited with single lobe of lungs.

By this report, we aimed to discuss the diagnosis and management of the case of bilateral type 1 congenital cystic adenomatoid malformation accompanied by hydrops fetalis diagnosed at second trimester.

Case Report

A 27-year-old, gravida 4, parity 2, abort 1 patient was referred to our clinic with an initial diagnosis of hydrops fetalis at 25+5 weeks gestation no medical and obstetric problem was found in her history. The biparietal diameter of the fetus was measured 65 mm, head circumference 254 mm, femur length 46 mm, abdominal circumference 342 mm (>97 percentile) and amniotic fluid index 200 mm during ultrasonographic examination. It was detected during fetus examination that there were multilocular anechoic cystic lesions (**Figs. 1-3**) which were maximum 26x24 mm, that cardiac axis shifted to left (85 degrees), and common ascites (**Fig. 4**) as well as skin edema and hydrocele were present. No additional anomaly was found during the examination of other systems. The pre-diagnosis of bilateral type 1 congenital cystic adenomatoid malformation accompanied by hydrops fetalis was established. The family was informed in terms of examination findings of fetus and the prognosis and it was offered to terminate the pregnancy. The consent of the patient was taken. Intracardiac potassium was applied to fetus. By inducting 400 µg intravaginal misoprostol application, 1650 gram male fetus in hydropic view was delivered with negative heartbeat. Hydrops fetalis and CCAM diagnoses were confirmed in postmortem pathology examination. No additional histopathological diagnosis was detected.

Discussion

The frequency of congenital cystic adenoid malformation is 1/11.000 – 35.000, and 80-95% of cases are unilateral and associated with single lobe of lungs.[8,9] CCAM is diagnosed as echogenic lesions in lungs which

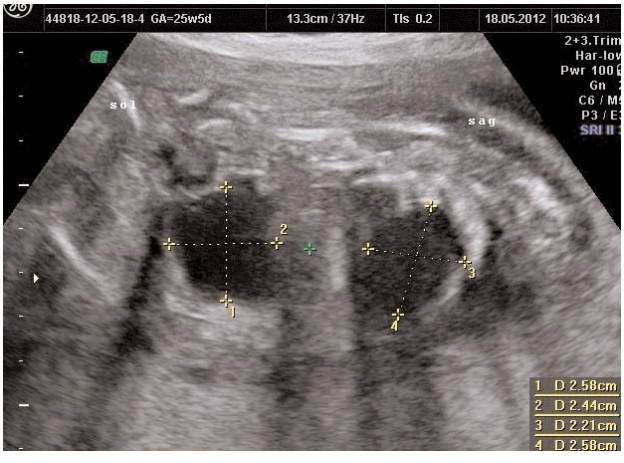

Fig. 1. Bilateral type 1 CCAM view on transverse section.

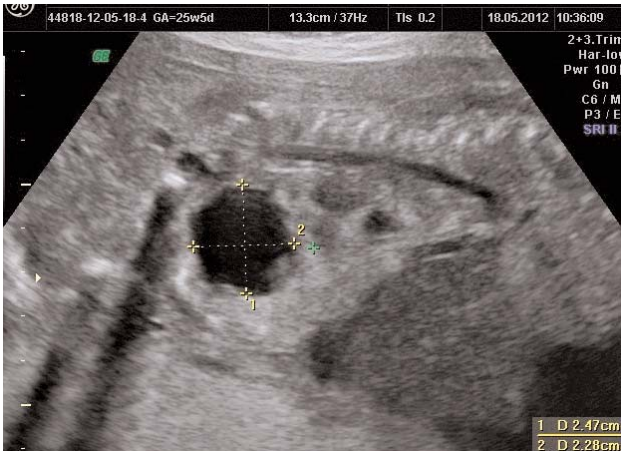

Fig. 2. Type 1 CCAM view in right lung on sagittal section.

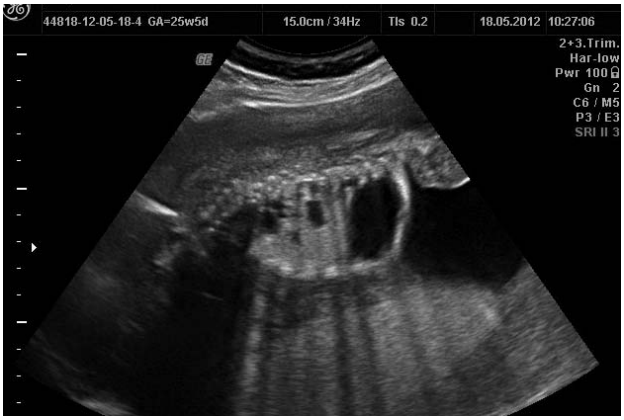

Fig. 3. Type 1 CCAM view in left lung on sagittal section.

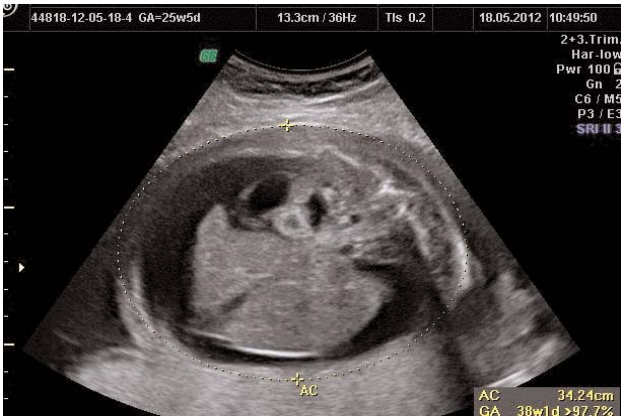

Fig. 4. The view of ascites on transverse section.

are solid, cystic or including both forms during ultrasonographic examination at second trimester.[8-11] Gornall et al. reported that the accuracy of ultrasonography for CCAM at prenatal period is 81% and the positive predictive value is 57%.[9] In order to decrease the error rate in prenatal diagnosis of CCAM, differential diagnosis for diaphragmatic hernia, extralobar and intralobar bronchopulmonary sequestration, lobar emphysema, bronchogenic cyst, and mediastinal lesions such as cystic teratoma or neurenteric cyst.[8-12] In differential diagnosis, Doppler ultrasonography and especially fetal MRI during advanced gestational weeks are helpful. The association of CCAM with bronchopulmonary sequestration, tracheal obstruction, bronchial atresia and lobar emphysema is not rare.[10]

Illanes et al. conducted a study including 43 CCAM cases and found out that 56% of cases had macrocystic lesions (>5 mm), 67% of them had mediastinal shift, and 19% of them had hydrops fetalis. Also, mortality rate of cases developing hydrops was reported as 75%.[10] Ierullo et al. examined 34 CCAM cases and they reported that 20.6% of the cases were macrocystic and all of them were unilateral, 79.4% of them had mediastinal shift and hydrops developed in 17.6% of them.[11] In our case, we detected that there were macrocystic multiocular lesions in bilateral lungs, cardiac axis shifted to left and there was hydrops.

Calvert et al. also found out that their all CCAM cases were unilateral and hydrops developed in 8.7% of them. In 48% of CCAM cases had regression during antenatal period and also 8.7% of the cases had complete regression.[13] CCAM is isolated and it rarely associates with other structural anomalies. However, the association of type 2 CCAM cases with cardiac (truncus arteriosus and Fallot's tetralogy) or renal anomalies, gastrointestinal system atresias and skeletal dysplasias.[8-14]

The relationship of CCAM cases with chromosomal anomalies is not known. Follow-up and management of CCAM cases should be planned according to gestational week, CCAM volume (calculated by the formula of height x length x width x 0.52 for lesion after ultrasonographic image of CCAM is obtained) or CVR rate (CCAM volume/head circumference). If CVR is higher than 1.6, then fetal hydrops risk is high and it is recommended to carry out fetal evaluation three times a week; if it is lower than 1.2, then it is recommended to carry out fetal evaluation once a week.[16-18] It was reported that CRV displayed a rapid increase between 20 and 25 weeks gestation and decrease after 25 weeks gestation, and it was highlighted that follow-ups during these weeks are significant.[16]

The presence of mediastinal shift, amniotic fluid index, umbilical artery Doppler flow pattern, ductus venosus Doppler flow pattern and placental thickness are used as the other significant parameters in fetal follow-up. Prognosis is generally better in type 1 CCAM cases, but the prognosis is poor in the presence of hydrops fetalis, ascites, polyhydramniosis, bilateral lung involvement and mediastinal shift.[8-13,15] Isolated cases not accompanied by hydrops should be evaluated by follow-up parameters once every three weeks.[8] In cases with CCAM including single or multiple major cystic lesions and accompanied by hydrops cases, it was shown that thoracoamniotic shunt is useful if gestational week

is lower than 32 after the presence of other structural and chromosomal anomalies is excluded. However, it was reported that thoracocentesis is not effective and cystic substance accumulated again in a short time. Thoracoamniotic shunt is not recommended in the presence of multicystic or semisolid or weighted solid lesions. It is suggested to plan delivery after 32 weeks gestation.[17,18]

In our case, thoracoamniotic shunt was not considered due to the presence of multiple cystic lesions in bilateral lungs and family discord. Upon the request of family, pregnancy was terminated with the diagnoses of bilateral type 1 CCAM and hydrops fetalis.

Conclusion

The differential diagnosis of type 1 congenital cystic adenomatoid malformation cases and presence of other accompanying anomalies are significant in terms of management. Prognosis is better in isolated type 1 congenital cystic adenomatioid malformation cases, and they can be followed up conservatively. In cases of multicystic bilateral type 1 congenital cystic adenomatoid malformation, prognosis is poor in the presence of hydrops fetalis, ascites, polyhydramniosis and mediastinal shift, and termination of pregnancy can be recommended.

Conflicts of Interest: No conflicts declared.

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