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Congenital cystic adenomatous malformation: a case report and a literature review

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Background. A congenital cystic adenomatoid malformation (CCAM) is a foetal pulmonary development abnormality caused by airway dysgenesis that is characterized by cystic or adenomatous lesions in the terminal bronchioles. The size of the mass, the degree of the mediastinal shift, and the presence of hydrops and polyhydramnios can all affect the severity of a case. Treatment can be initiated at early stages by applying prenatal and postnatal methods. Because CCAM is a rare pathology that is often only accidentally diagnosed during routine ultrasounds, we would like to share our case report to enrich the literature on this pathology and to present a case successfully treated at our hospital.

Materials and methods. A patient with her first multiple pregnancy was seen for prenatal care and her first ultrasound at 17 weeks of gestation. One of the twins was diagnosed with a congenital cystic adenomatoid malformation of the left lung. At 20 weeks of gestation, an enlarged left lung with small cysts, a compressed right lung, a compressed and displaced heart, and oligohydramnios were observed. At 28 weeks of gestation, a fetoplacental circulation disorder appeared. At 32 weeks of gestation, due the unstable condition of the affected foetus, the twins were delivered via a C-section. The treatment of the newborn included antibiotics, caffeine citrate, and breathing therapy.

Results and conclusions. CCAM are often diagnosed by accident when performing routine pregnancy ultrasound examinations. CT is the most reliable X-ray-based examination method for confirming a diagnosis. When CCAM is suspected in the foetus, amniocentesis and cariotype identification are performed, but chromosomal anomalies related to CCAM are often not identified. Currently, the best treatment results have been achieved by applying combined prenatal therapy and early surgical treatment.

Keywords: congenital cystic adenomatoid malformation, multiple pregnancy, dichorionic twins, prenatal diagnosis

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INTRODUCTION

Congenital cystic adenomatoid malformation (CCAM), also known as congenital cystic degeneration, is a rare abnormality of foetal lung development. This disease is characterized by benign cystic or adenomatoid lung tumours that grow on the terminal bronchioles and impair their function. It is caused by impaired development of the respiratory tract. According to various research data, the incidence of CCAMs is reportedly between 1:8300 and 1:35000 live births (1). Even though this malformation more commonly affects males, there is no evidence that sex has any effect on the probability of the development of this disease in individuals (2, 3). In this article, we present a CCAM case report for a female patient.

CASE REPORT

A 26-year-old woman with her first multiple pregnancy was seen for prenatal care and her first ultrasound at 17 weeks of gestation. One of female dichorionic diamnionic twins was diagnosed with a congenital cystic adenomatoid malformation of the left lung. In order to determine the possibility of a genetic origin of the disease, tests of the cariotype of the affected foetus, of the 13th, 18th, and 21st chromosomes of the healthy foetus, and of gonosomal aneuploidy were performed. There were no genetic disorders found in either foetus. At 20 weeks of gestation, a second ultrasound was performed. An enlarged left lung with small cysts (Fig. 1), a compressed right lung, a compressed and displaced heart, and oligohydramnios were observed (Fig. 2). Recommendations included: consultation to discuss further prenatal and delivery practices, ultrasound examinations every 3–4 weeks (while monitoring the amniotic fluid levels and the condition of both foetuses), consultations with a paediatric surgeon, and the delivery of the twins in a hospital specializing in high-risk and complicated pregnancies.

There were no significant changes in the condition of the affected foetus in subsequent ultrasound examinations. At 28 weeks and 4 days of gestation, a dopplerometric examination revealed a fetoplacental circulation disorder in the affected foetus with increased PI (Pulsatility Index), RI (Resistance Index), and S/D indexes.

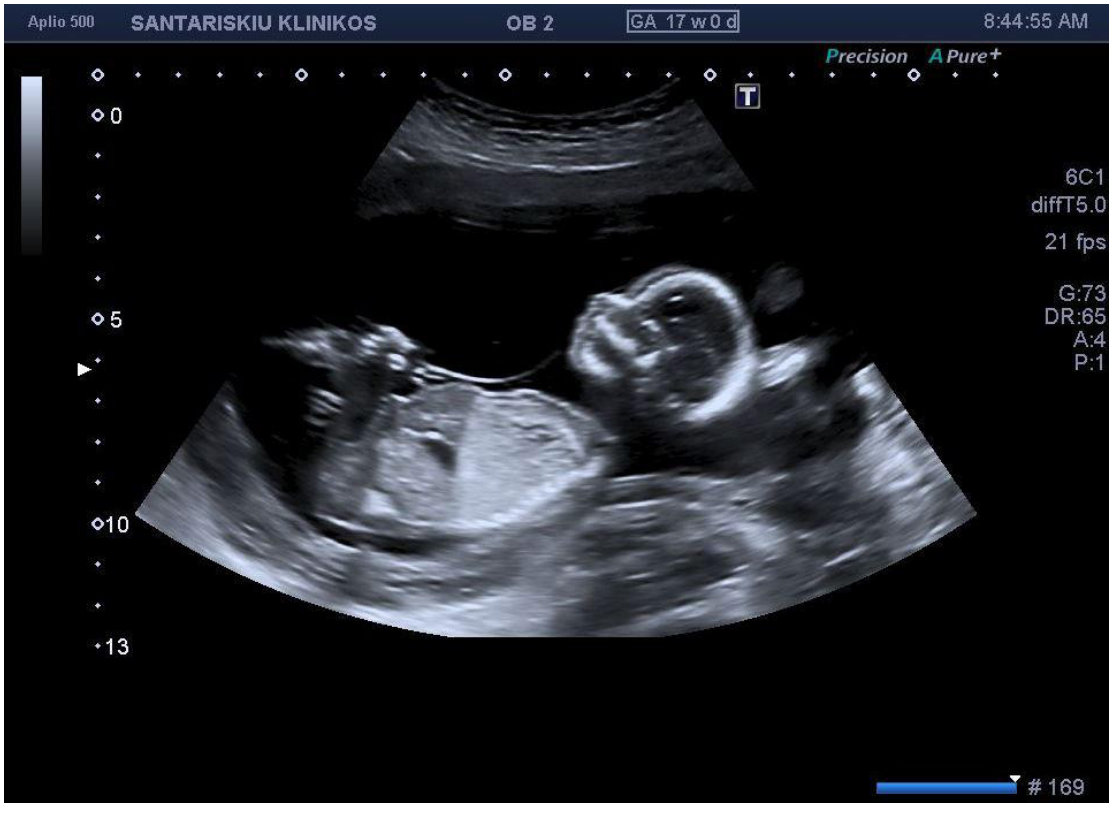

Fig. 1. 17 weeks of gestation. Twin B. An enlarged left lung with small cysts

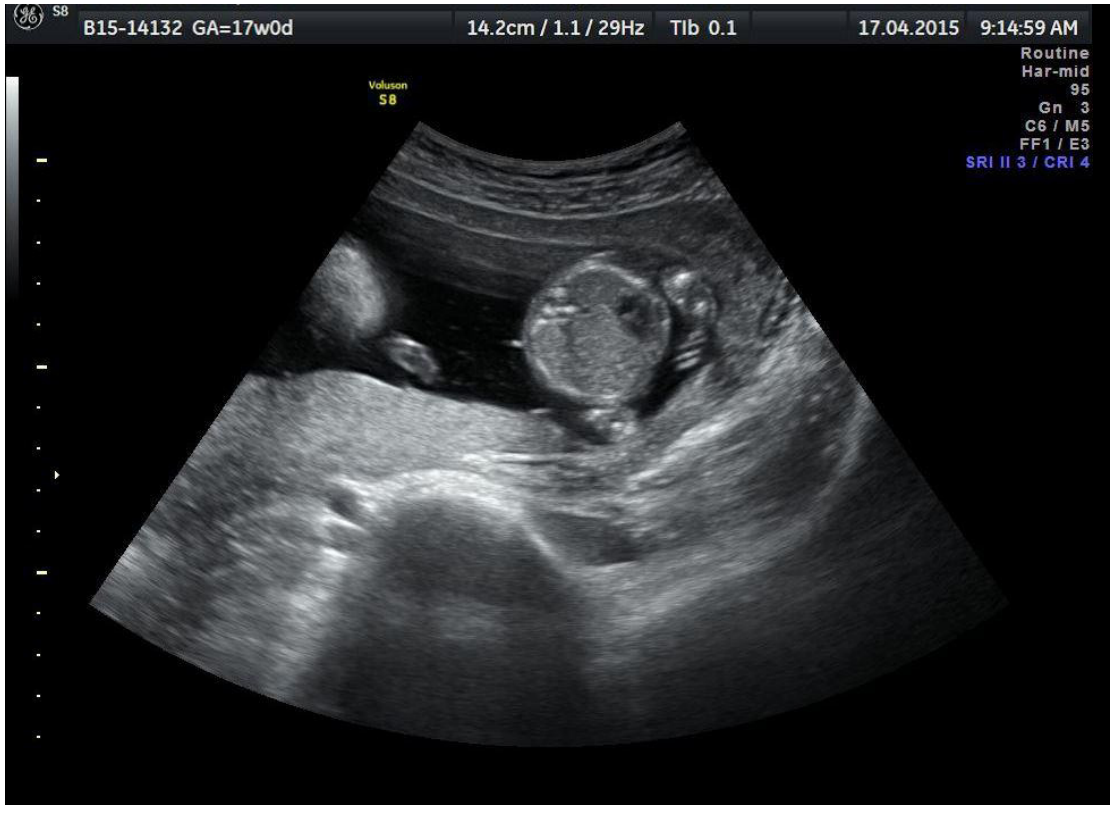

Fig. 2. 17 weeks of gestation. Twin B. The compressed and displaced heart

At 30 weeks of gestation, the pregnant woman came to the emergency room complaining of lower abdominal cramps. A false labour was suspected and an ultrasound showed a 14% difference in growth between the foetuses. Recommendations included outpatient care, a resting regime, monitoring of the foetuses' movements, and hydration (two litres or more per day). The woman was instructed to come to the emergency room immediately in the event of any vaginal blood discharge, reductions in foetal movements, regular labour contractions, or if her waters broke. Another consultation was scheduled 10–14 days later at the Perinatological Coordination Centre of Vilnius University Hospital.

At 32 weeks and 4 days of gestation, the lower abdominal cramps returned with no reduction of foetal movements. Due to unequal foetal growth, the unstable condition of the affected foetus and the fetoplacental circulation disorder, it was decided to perform a pulmonary maturation and deliver the twins.

Due to the unstable condition of the diseased twin, a C-section was performed. The weight and height of the first female newborn at birth was 1870 g and 45 cm, with Apgar scores of 8/8 at 1 and 5 minutes after birth. The weight and height of the second female newborn at birth was 1320 g and 37 cm, with Apgar scores of 7/8 at 1 and 5 minutes after birth. CPAP therapy was started shortly after birth for both newborns due to progressive respiratory failure, impaired microcirculation, their need for oxygen, and their prematurity. A bell-shaped symmetrical chest, weak breathing sounds in both lungs, and intense movement of the intercostal spaces while breathing were observed in the second newborn. A chest X-ray showed reduced aeration in the left lung, a sharper X-ray image of the right lung, weak differentiation of the left diaphragm vault, and the displacement of the mediastinal organs to the right. The diagnosis was a congenital cystic adenomatosis of the left lung (Q33.0). Because a surgical pulmonary disease was suspected in the second twin, both newborns were transferred to the Neonatal Intensive Care Unit (NICU) at the Neonatology Centre of the Children's Hospital. The placenta and umbilical cords were histologically tested and no inflammation was observed.

After transferring the newborn to the NICU, her condition was assessed as very critical due to the aforementioned symptoms of respiratory dysfunction. High-frequency oscillating ventilation with an oxygen concentration of 21% was begun. Her skin was marble-coloured with a capillary refill time (CRT) of 4–5 s, the heart was functioning rhythmically, the tones could be heard better on the right side of her thoracic cavity, her pulse was 100–170 bpm, and saturation was 72–92% with a 50% supply of O_2. The activity of her nervous system was reduced and she was hypotonic.

General, biochemical (CRP, electrolytes, glucose) and blood gas tests were performed. Based on the results of these tests, respiratory acidosis was identified: pCO_2 – 40.7 mmHg, HCO_3 – 22.2 mmol/L, SBE – –2.5. The following changes were observed after performing ultrasound and X-ray examinations of the thoracic cavity: non-homogenous, partially hepatised tissue with multiple cysts and blood flow in the left pulmonary area, and uneven aeration of the left lung. A computer tomography (CT) test revealed compensationally increased aeration of the superior and anterior parts of the inferior lobe of the left lung; heterogeneously dense masses with blood flow from the aorta in the inferior part of the cardiodiaphragmal corner and paravertebrally; a slight dislocation of the mediastinum to the right.

Despite prematurity and a critical condition, there were no indications for a biopsy or an urgent surgical intervention. Treatment was prescribed for the newborn: penicillin (7 days), gentamicin (7 days), and caffeine citrate (8 days). The newborn's condition stabilized over time and her pH balance normalized. As her condition improved, CPAP therapy was applied on the 5th day of her life. Her breathing therapy ended on the 11th day of her life.

After 12 days of treatment at the NICU, the newborn was transferred to the Premature Newborn Division, where she was treated with caffeine citrate, vitamin D, and iron supplements. Due to her good condition and growing weight, the newborn was discharged at the age of 36 days. At discharge, regular follow-up of the newborn's development and health on an ambulatory basis was recommended.

DISCUSSION (REVIEW OF THE LITERATURE)

Diagnostics

CCAM pathology is often diagnosed by accident when performing routine pregnancy ultrasound examinations that allow for the observation of non-specified multiple large (type I) or small (type II cysts (Fig. 2). In the case of type III, the especially small size of the cysts causes ultrasound examinations to reveal homogenous masses (2, 4).

By observing one-sided cystic pulmonary growths, CCAM can be differentiated from congenital diaphragmatic hernias, oesophageal duplication (doubling), and congenital lobe emphysema. If the CCAM is seen as a solid mass during the ultrasound examination, it should be separated from a pulmonary sequestration (5).

Magnetic resonance imaging (MRI) is an equally informative examination method for pregnant women, which helps to diagnose the development of a pulmonary anomaly and to differentiate it from other conditions, especially from congenital diaphragm hernias. In addition, in the cases of obese pregnant women, incorrect foetal positioning, and oligohydramnios, MRIs are often a better examination method than ultrasound tests (2).

In an thoracic cavity overview X-ray, the most frequent indication of CCAM is the tissue formed of air-filled cysts. Other typical non-specified indications can include: mediastinum dislocation, exudate in the pleural and/or pericardial spaces, or pneumothorax (2).

CT is the most reliable X-ray-based examination method for confirming a CCAM diagnosis. A typical sign is thin-walled and liquid-air-surfaced multifocal cystic growths surrounded by normal pulmonary parenchyma. For more accurate differentiation and typing, high-resolution CT scan can be performed.

When CCAM is suspected in the foetus, amniocentesis and cariotype identification are performed, but chromosomal anomalies related to CCAM are often not identified, as was the case in the clinical case described.

Risks for the foetus and the outcomes

In certain cases, when a macrocystic form of CCAM appears, there is a risk of pulmonary hypoplasia and the development of foetal hydrops

due to the compression of the developing lungs by the large cysts (5).

Foetuses suffering from CCAM can have various survivability prognoses. They depend on the following factors: the size of the growth, the degree of mediastinal displacement, hydrops and polyhydramnios. Good, moderate, and poor prognosis groups can be identified based on these factors. However, as the number of studies examining CCAM is growing around the world, it is being discovered that many cystic growths regress on their own in the third trimester of pregnancy (5).

Treatment

Despite the fact that CCAM is a rare and poorly researched anomaly, data from various literature sources suggest that treatment can be started in the initial stages of the pathology development (6). A number of treatment methods have currently been developed around the world that can be applied pre- and postnatally.

Prenatal treatment methods include cystic growth aspiration or draining, thoracoamniotic bypass, the injection of sclerosing agents, and foetal surgery (7, 8). Combined prenatal treatment is frequently applied, one of the main components of which is thoracoamniotic bypass. The method is based on the principle of constant drainage of the cyst-filling fluid into the amnion sac (2). Macrocystic pulmonary growths are usually bypassed. Research performed to date confirms the effectiveness of bypass when used to avoid pulmonary hypoplasia and the fetal hydrops related to it (9). If there is no opportunity to perform a bypass, the first-choice alternative is surgical treatment (loboectomy). The successful removal of the anomalous growth provides the right conditions for the continued growth and development of the healthy part of the lung. Another alternative to bypass is cystic sclerosation, which is most effective when there is pleural exudate, cystic hygroma and bronchopulmonary sequestration (7).

The continuation of treatment of the foetus after birth is surgery aimed at prevention of CCAM complications (pneumothorax, recurring infections, malignancy) (10). To achieve optimal results, it is recommended to perform the surgery by the age of 12 months. If the affected part of the lung is not removed, there is a chance that it may develop into a pulmonary rabdomyosarcoma in late childhood.

Indications for surgical treatment were not observed in the clinical case described, so a conservative treatment method based on antibiotic and oxygen therapies was chosen.

CONCLUSIONS

CCAM is a benign cystic or adenomatous terminal bronchiole disorder caused by a respiratory tract genesis disorder. Its prenatal diagnosis is confirmed based on MRI data and its postnatal diagnosis is confirmed by indications observed in CT, though its early diagnosis is based on ultrasound examinations of the pregnant woman. Currently, the best treatment results have been achieved by applying a combined prenatal therapy, especially with thoracoamniotic bypass for the foetus, and early surgical treatment for the newborn. In the near future, the greatest successes in CCAM treatment are expected from improving foetal surgical methods.

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ĮGIMTA PLAUČIŲ CISTINĖ DEGENERACIJA: KLINIKINIO ATVEJO PRISTATYMAS IR LITERATŪROS APŽVALGA

Santrauka

Tikslas. Įgimta plaučių cistinė degeneracija (CCAM) – tai reta vaisiaus plaučių vystymosi patologija. Šiai ligai būdingas gerybinis cistinis ar adenomatozinis terminalinių bronchiolių pažeidimas, kurį nulemia kvėpavimo takų genezės sutrikimas. Vaisių išgyvenamumo prognozė priklauso nuo darinio dydžio, tarpuplaučio poslinkio laipsnio, vandenės, polihidramniono. CCAM gydymas gali būti pradėtas jau ankstyvose patologijos vystymosi stadijose taikant pre- ir postnatalinius metodus. Kadangi CCAM yra reta ir dažniausiai atsitiktinai ultragarsu nustatoma patologija, ją aptarsime ir pristatysime mūsų ligoninėje sėkmingai gydytą atvejį.

Medžiaga ir metodai. Pateikiame įgimtos plaučių cistinės degeneracijos atvejį. Pacientė pirmą kartą apsilankė prenatalinei priežiūrai ir ultragarsiniam tyrimui 17-ąją savo pirmojo daugiavaisio nėštumo savaitę. Vienam iš dichorioninių diamnioninių dvynių buvo diagnozuota kairiojo plaučio įgimta cistinė degeneracija. 20-ąją nėštumo savaitę ultragarsinio tyrimo metu buvo aptikta: padidėjęs kairysis plautis, kuriame gausu nedidelių cistų, suspaustas dešinysis plautis, suspausta ir dislokuota širdis bei oligohidramnionas. 28-ąją savaitę diagnuozuotas fetoplacentinės kraujotakos sutrikimas. 32-ąją savaitę dėl nestabilios sergančio vaisiaus būklės nuspręsta užbaigti nėštumą. Po gimimo naujagimiui buvo taikytas gydymas antibiotikais, kofeino citratu ir kvėpuojamąja terapija.

Rezultatai ir išvados. CCAM neretai diagnozuojama atsitiktinai, vaisių tiriant ultragarsu. Patikimiausias diagnozę patvirtinantis tyrimas – kompiuterinė tomografija. Įtariant minėtą patologiją, rekomenduojama atlikti amniocentezę ir kariotipo nustatymo tyrimą, tačiau neretai jokių chromosominių pakitimų, siejamų su CCAM, nerandama. Šiuo metu geriausi gydymo rezultatai pasiekiami derinant prenatalinę terapiją ir ankstyvą chirurginį gydymą. Tikimasi, kad ateityje ši liga bus gydoma operuojant vaisių dar esantį gimdoje.

Raktažodžiai: įgimta cistinė plaučių degeneracija, daugiavaisis nėštumas, dichorioniniai dvyniai, prenatalinė diagnostika