Case Report

Congenital Cystic Adenomatoid Malformation Type III Associated With Congenital Anomalies

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SUMMARY

CONGENITAL CYSTIC ADENOMATOİD MALFORMATION TYPE III ASSOCIATED WITH CONGENİTAL ANOMALIES

Background: We present a case of congenital cystic adenomatoid malformation of the lung, type III, associated with congenital anomalies.

Observation: Still born male fetus, whose karyotype found to be 46XY, was delivered to a 22 year old white female at 19th weeks of gestation. The parents of the baby were first degree relatives. During the follow-up of the pregnant a fetus with lomber bifid spine and ascites was observed. Clinical prediagnosis was cystic fibrosis.Upon examination of the internal organs, both of the lungs were found to be firm, bulky and had depressions of the ribs on their surfaces due to thoracic cage pressure. Cross sections showed a few scattered cysts 0.1-0.2 mm in diameter. Microscopic examination revealed alveolus-like structures lined by low cuboidal epithelium separated by loose masenchymal interstitial tissue and some bronchiole-like structures spread in between. Additionally, cartilage and mucogenic cells, as well as elastic tissue and smooth or striated muscle fibers were not detected around these structures.

Conclusion: With all these data in hand, our case is an example of Congenital Cystic Adenomatoid Malformation of the Lung, Stocker type III.

Key Words: Congenital cystic adenomatoid malformation (CCAM), Congenital pulmonary airway malformation (CPAM)

ÖZET

KONJENİTAL ANOMALİLERİN EŞLİK ETTİĞİ BİR KONJENİTAL KİSTİK ADENOMATÖZ MALFORMASYON TİP III OLGUSU

Amaç: Konjenital anomalilerin eşlik ettiği akciğere ait konjenital kistik adenomatöz malformasyon tip III olgusu sunuldu.

Olgu: Karyotipi 46 XY olan ölü erkek bebek, gebeliğin 19. haftasındaki düşüğü takiben incelendi. Anne 22 yaşında olup eşi ile birinci dereceden akraba idi. Takipler sırasında fetusta asit ve spina bifida saptanmış ve kistik fibrozdan şüphe edilmişti. Otopside her iki akciğer sert ve büyük olup torasik basınca bağlı olarak kaburgaların izlerini taşımaktaydı. Kesitlerde birbirinden ayrı duran 0.1-0.2 mm çapında kistler mevcuttu. Mikroskopik incelemede, içinde bronşiole benzer yapılar bulunan gevşek bir mezankimal interstisyum ile birbirlerinden ayrılan, alçak kübik epitel ile döşeli alveol benzeri yapılar saptandı. Ayrıca bu yapıların çevresinde ne kıkırdak doku ve mukojen hücreler, ne de elastik doku ile düz veya çizgili kasa ait yapılara rastlandı.

Sonuç: Bu bulgularla olguya akciğerin konjenital kistik adenomatoid malformasyonu, Stocker tip III tanısı koyuldu.

Anahtar Kelimeler: Konjenital kistik adenomatoid malformasyon, Konjenital pulmoner traktus malformasyonu

Congenital cystic adenomatoid malformation of the lung (CCAM) is a rare disorder mostly seen in newborn and stillborn infants (1). It is a developmental malformation of the lungs in which there is a defect in formation of terminal and respiratory bronchioles and alveolar ducts (2).

Our case is a bilateral CCAM, type III associated with congenital anomalies. Because of the rarity of bilaterality in CCAM cases and association of congenital anomalies in CCAM type III, together with this case being the only one amongst our autopsies; we have found it worthwhile to report.

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Figure 1. *Gross view of the fetus: stillborn male fetus at 19 week of gestation.*
External abronmalities: Slightly large head, low set ears, fusion of the eyelidis with globes properly placed, wide and depressed nasal root, chin smaller than normal, syndactyly, distended abdomen due to fetal ascites, lomber spina bifida, imperforeted anus and maceration observed.

Figure 2. *Grossly both of the lungs are firm and bulky, showing depressions on the surface due to thoracic cage pressure. A few scattered small cysts 0,1-0.2 mm in diameter seen both on the surface and in the cross section of the lungs.*

CASE

Our case is a stillborn male fetus weighning 250 gr, at 19th week of gestation, delivered to a 22-year old white female prima gravid. Mother, married her 1st degree relative (her uncle's son), smoked 5 cigarettes a day. During her follow-up in a non-university hospital, two anomalies, lomber spinabifida and fetal ascites were observed in USG and pancreatic cystic fibrosis was suspected. Upon tissue culture karyotype 46XY was determined.

AUTOPSY FINDINGS

Gross autopsy examination revealed the following features: Macera male fetus with a slightly large head, low set ears with depression of the nasal root, fused eyelids with globes in proper position, syndactyly, chin smaller than normal, distended abdomen due to ascites, lomber bifid spine and imperforation of the anus (Figure 1).

In internal examination, grossly both of the lungs were firm, bulky and showed depressions on the surfaces due to thoracic cage pressure (Figure 2).

A few scattered small cysts 0.1-0.2 in mm diameter were observed both on the surface and the cross section of the lungs. Also there was a hematoma within the heart chambers, and ascites in the abdomen. No other abnormalities in internal or external organs were detected.

Upon microscopic examination the malformation was seen to consist of alveolus-like structures lined by low cuboidal epithelium separated by loose mesenchymal interstitial tissue and interspersed bronchiole-like structures (Figure 3A-3B).

Cartilage and mucogenic cells were not detected No elastic tissue or smooth muscle were found around these structures. Also, no striated muscle were seen in the mesenchymal tissue. With these findings, our case demonstrates the pathological features of congenital cystic adenomatoid malformation of the lung, Stocker type III.

DISCUSSION

Previously, the term "congenital cystic disease of the lung" was used for a wide variety of cystic pulmonary abnormalities upon detection of radiolucencies in chest roentgenograms without pathologic confirmation. A study by Koontz showed that some cases of cystic fibrosis, postinflammatory pneumatocels and bronchiectasis as well as some sequestrations, hilar and bronchogenic cysts, and emphysematous lesions were reported as congenital cystic disease of the lung (5).

In 1949, separate entity, "congenital adenomatoid malformation of the lung" was designated by Chin and Tang (3). CCAM is a developmental mal-

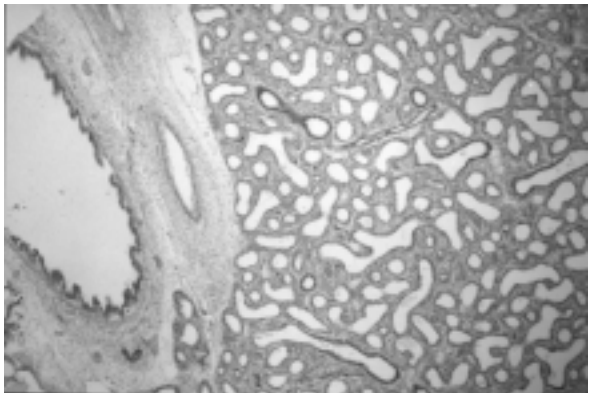

Figure 3A. *Fetal microscopic findings: Low-power view of the lung: A portion of the bronchus can be seen on the right and the multiple bronchiolar/alveolar duct like structures (resembling an immature lung) (HEX40).*

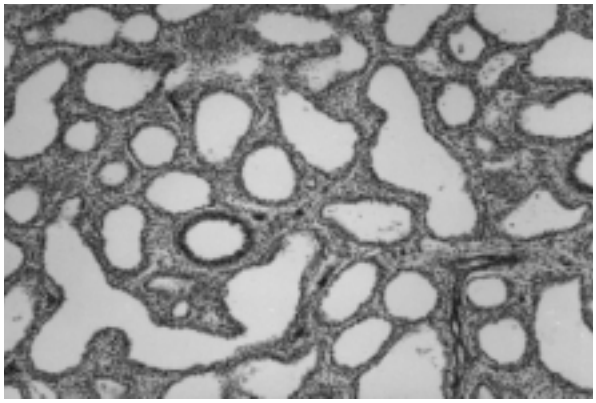

Figure 3B. *High power view of the lung: Microscopically, Bronchiolar/alveolus-like structures lined by low cuboidal epitelium surrounded by interstitial tissue mesenchymal in character (Type 3 CCAM) (HEX100).*

formation of terminal and respiratory bronchioles and alveolar ducts (2). Althoughs, CCAM is mainly a disorder of the newborn and stillborn infants, there have been 2 adult cases of CCAM reported in the literature (1, 4).

The lesions seen in CCAM are most of the time unilateral (confined to one lobe/ segment/ whole lung); bilateral lesions were very rarely detected (1,5). Here, in our case we have encountered bilateral lesions in the lungs.

CCAM is usually accompained with maternal polyhydramnions, hydrops fetalis and fetal ascites (1,5,6). Fetal ascites was present in the case we present.

On the other hand, congenital anomalies such as bilateral renal agenesis/dysgenesis, extralobar pulmonary sequestration, cardiovascular malformation, diaphragmatic hernia, jejunal atresia, pulmonary hypoplasia and skeletal malformations may be associated with this entity; most commonly in type II (3, 5, 6). Our case, CCAM type III, was also associated with congenital anomalies.

Recently, a new attempt has been made by Dr. Thomas Stocker, MD, to rename this entity as "Congenital pulmonary airway malformation", for CCAM is a defect in differentiation of the tracheobronchial tree. According to Dr. Stocker, the classification of CCAM falls into 5 categories (7), which are:

A. CCAM Type O (CPAM): Acinar dysgenesis (agenesis)-Tracheobronchial origin.

B. CCAM Type 1 (CPAM 1): Large cyst type of bronchial/bronchiolar origin

C. CCAM Type 2 (CPAM 2): Intermediate cyst type of bronchiolar origin

D. CCAM Type 3 (CPAM): Small cyst type of bronchiolar/alveolar duct origin

CCAM Type 3 which almost exclusively occurs in males (as is the case we present), accounts for 8-10% of CCAM cases. In 79% of the cases it is associated with maternal polyhydramnios. This type of CCAM is the original congenital adenomatoid malformation of theğ lung described by Chin and Tang in 1949 (3).

Grossly, harge bulky lesions involve the entire lobe or even an entire lung. In our case the lesions entirely involved both of the lungs.

Microscopically, lesions consist of randomly scattered bronchiolar/alveolar duct-like structures lined by low cuboidal epithelium, which are surrounded by alveoli lined by cuboidal epithelium (Figures 3A, 3B, 4A, 4B).

E. Type 4 (CPAM 4): Peripheral cyst type distal acinar origin.

In our 10-year series of autopsies we have had only a few CCAM case, with this case being the only one associated with congenital anomalies.

Briefly, we present a case of bilateral CCAM type 3 associated with congenital anomalies, diagnosed in the 2nd trimester as a still fetus.

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