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A Case of Mesenchymal Tumor Developing on the Background of Congenital Pulmonary Airway Malformation

Konjenital Pulmoner Havayolu Malformasyonu Zemininde Gelişen Mezenkimal Tümör Olgusu

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Abstract

Congenital pulmonary airway malformations are rare developmental lung anomalies, five types of which have been classified by Stocker. The condition is generally diagnosed prenatally during routine prenatal ultrasonography, however, some cases are asymptomatic and so may not be diagnosed until later in life. Clinically, the condition can present in newborns with shortness of breath, cyanosis and respiratory distress, and may lead to recurrent infections in later ages, and malignant transformations have been defined in literature. Here, we present a case that may be of interest to literature not only due to the diagnosis later in life, but also due to the development of a mesenchymal tumor in the background.

Keywords: *Congenital pulmonary airway malformation, Mesenchymal Tumor, Bronchopulmonary Sequestration.*

Öz

Konjenital pulmoner havayolu malformasyonu nadir görülen ve akciğerin gelişimsel bir anomalisidir. Stocker tarafından sınıflandırılmış olup beş tipe ayrılmıştır. Genellikle, prenatal rutin ultrasonografi sayesinde tanı bu dönemde konur. Ancak bazı olgular asemptomatik seyrettiği için ileri yaşa kadar tanı almayabilir. Klinik olarak, nefes darlığı, siyanoz ve yenidoğan döneminde respiratuar distress ile prezente olabilir. İleri yaşta tekrarlayan enfeksiyonların nedeni olabilir. Malign transformasyon literatürde tanımlanmış bir antitedir. Olgumuz hem ileri yaşta tanı almış olması hem de mezenkimal tümör gelişimi nedeniyle literatüre katkı sağlamak için sunuldu.

Anahtar Kelimeler: *Konjenital Pulmoner Havayolu Malformasyonu, Mezenkimal Tümör, Bronkopulmoner Sekestrasyon.*

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Congenital pulmonary airway malformations (CPAM), formerly known as congenital cystic adenomatoid malformations, are developmental lung anomalies that are generally diagnosed prenatally during routine ultrasonography scans, although diagnoses may be made incidentally or secondary to recurring infections at later ages. Bronchopulmonary sequestration, bronchogenic cyst and congenital diaphragmatic hernia should be considered in the differential diagnosis of CPAM. We consider this case to be of interest to literature due not only to the later-life diagnosis, but also the mesenchymal tumor that developed in the background of the patient's pre-existing disease.

CASE

A 52-year-old female patient with essential thrombocythemia and hyperthyroidism was referred to our clinic with clubbed fingers and shortness of breath. We were informed of a history of long-term shortness of breath and a cough for the last five months, although the patient reported no phlegm, hemoptysis or chest pain. The patient had a 30-pack/year smoking history but had quit 5 years earlier. SpO_2 was 97% in room air. A respiratory system examination revealed drastically decreased respiratory sounds in the right lung, and an analysis of the patient's medical records revealed that the results of a thoracic computed tomography (CT) performed in previous years indicated cystic adenomatoid malformation (Figure 1). A new tomography scan was carried out, and a pulmonary function test was performed due to the increase in symptoms at that time. An obvious restriction was identified in a pulmonary function test (PFT): FEV1/FVC: 75%, FEV1: 0.99 L (38% Pred), FVC: 1.33 L (40% Pred), while a thoracic CT revealed a mass lesion with calcific components on the right hemithorax (Figure 2). The patient was referred to the interventional radiology department, where a Tru-Cut biopsy was performed on the lesion revealing a spindle cell mesenchymal mass lesion. PET-CT revealed a slightly increased FDG uptake and an SUVmax value of 3 in the mass, which measured 183 × 173 × 215 mm in size with calcification to the wall and partial cystic necrotic areas in the lower lobe of the right hemithorax, but no significant FDG uptake in extrapulmonary areas. The patient underwent surgery in a different medical facility during which a mass lesion weighing approximately 3.5 kg was removed (Figure 3). The pathology results showed mesenchymal tumor proliferation with hamartomatous/teratomatous components. The tumor's diameter, the presence of necrosis and the distinct proliferation of spindle cells across larger areas indicated low-grade sarcomatous development, and the patient was recommended for follow-up. A postoperative thoracic CT revealed the right lung to be fully opened other than the tissue defects associated with surgery and postoperative changes (Figure 4). PFT revealed FEV1/FVC: 81%, FEV1: 2.42 L (94% Pred), FVC: 3.00 L (92% Pred). The patient's shortness of breath improved considerably and did not worsen during follow-up, which is continuing at our center.

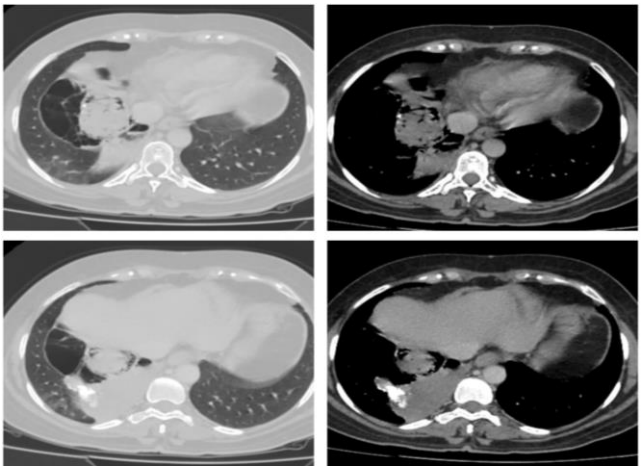

Figure 1: *Cystic adenomatoid malformation in the lower lobe of the right lung*

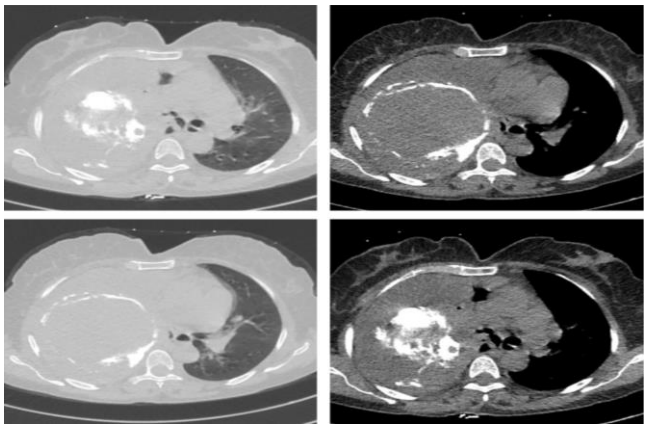

Figure 2: *Mass with calcific components in the right hemithorax*

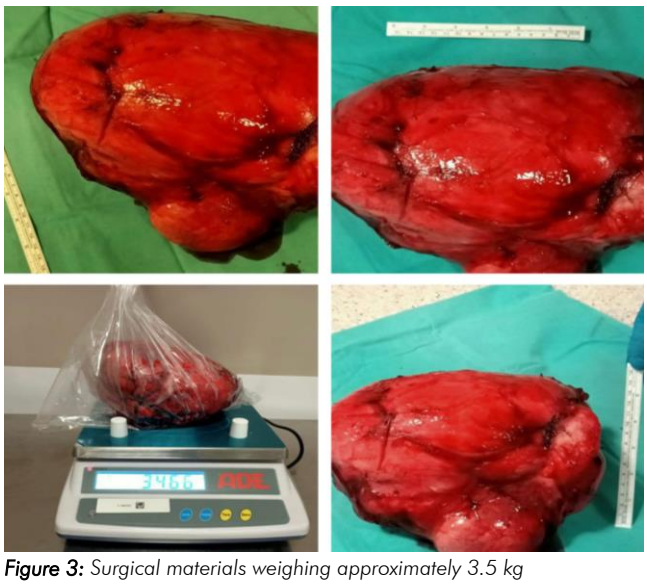

Figure 3: *Surgical materials weighing approximately 3.5 kg*

DISCUSSION

CPAM is a rare developmental lung anomaly that is seen in one in every 10,000–35,000 live births (1). It is generally diagnosed during prenatal scans in the 18th–20th weeks of pregnancy. Stocker put forward three classifications of congenital adenomatoid cystic malformations in 1977 (2) and added two further types in 2002 and updated the name of the disease to CPAM (3). It results from type 0 trachea or main bronchus. It is the most rarely observed type and is fatal. Type 1 is the most frequently encountered form, which develops in the distal and proximal bronchi, and accounts for 50–70% of all CPAM cases. Type 2 emerges from the terminal bronchioles and accounts for 15–30% of all cases, while type 3 originates in the alveoli and is considerably rare, developing as cysts that are microscopic in size and look like solid masses. Finally, type 4 also originates in the alveoli and presents multiple larger cysts.

A differential diagnosis of CPAM should consider bronchopulmonary sequestration, bronchogenic cysts and congenital diaphragmatic hernia, among which bronchopulmonary sequestration is most important, involving non-functional lung tissue fed by systemic circulation, and generally located in the lower lobes of the lung. Differentiating bronchopulmonary sequestration from CPAM can be challenging. Bronchogenic cysts occur with abnormal branching of the tracheobronchial tree. The least likely differential diagnosis, on the other hand, is congenital diaphragmatic hernia (4).

CPAM can be asymptomatic but can also present with shortness of breath, cyanosis and respiratory distress in newborns and infants, and can both be found incidentally in asymptomatic adults and as the source of recurring infections (5).

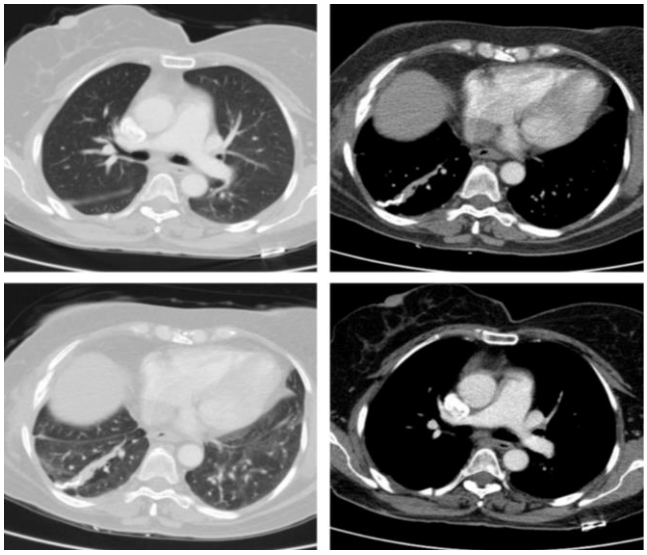

Figure 4: *Changes in sequela on postoperative thoracic CT*

The development of malignancy with CPAM has been defined in literature. Kaslovsky et al. (6) reported the development of bronchoalveolar carcinoma in a patient with type 1 CPAM who underwent an incomplete resection. Benouaich et al. (7) made a diagnosis of mixed bronchoalveolar and papillary adenocarcinoma after carrying out a surgical intervention on a 77-year-old patient with CPAM. Granata et al. (8) identified bronchoalveolar carcinoma in eight CPAM patients and rhabdomyosarcoma in five patients with CPAM. Our patient, who had long-term dyspnea and was diagnosed late, developed sarcomatous tumors, and her complaints improved significantly after surgery. Her follow-up is continuing.

We present this case to literature due to her development of a low-grade mesenchymal tumor that is rarely described in the literature.

CONFLICTS OF INTEREST

None declared.

AUTHOR CONTRIBUTIONS

Concept - T.K., Ş.I., H.S.Ş.; Planning and Design - T.K., Ş.I., H.S.Ş.; Supervision - T.K., Ş.I., H.S.Ş.; Funding - T.K., Ş.I., H.S.Ş.; Materials - T.K., Ş.I., H.S.Ş.; Data Collection and/or Processing - T.K., Ş.I., H.S.Ş.; Analysis and/or Interpretation - T.K., Ş.I., H.S.Ş.; Literature Review - T.K., Ş.I., H.S.Ş.; Writing - T.K., Ş.I., H.S.Ş.; Critical Review - T.K., Ş.I., H.S.Ş.

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