The European Research Journal 2023;9(6):1537-1540 Case Report

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Cystic fibrosis diagnosed in a nineteen-year-old case

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ABSTRACT

Cystic fibrosis is the most common autosomal recessive hereditary disease in white populations. It is characterized by the formation of abnormal secretions in the exocrine glands located in the sweat and salivary glands, tracheobronchial tree, large intestine, and pancreas. The severity of the clinic depends on the type of "cystic fibrosis transmembrane regulatory protein" gene mutation. Although most cases are diagnosed in infancy or childhood, some patients are also diagnosed during adolescence and adulthood. We report a case a 19-year-old patient who was followed up with a diagnosis of asthma and bronchiectasis since childhood and diagnosed with cystic fibrosis.

Keywords: Adolescence, bronchiectasis, cystic fibrosis

Cystic Fibrosis (CF) is the most common autosomal recessive (OR) inherited disease in the Caucasian race and its frequency is 1 in 2000 to 3000 live births. The main disorder is the formation of abnormal secretions in exocrine glands in sweat and salivary glands, tracheobronchial tree, large intestine, and pancreas [1].

The severity of CF clinic depends on the type of CFTR (Cystic fibrosis transmembrane regulatory protein) gene mutation. Symptoms and signs include recurrent pulmonary infections, pancreatic insufficiency, and high sweat chloride levels [1]. Since the CFTR protein can function, albeit partially, in some patients with CF, their clinics are mild and show atypical symptoms. While most of the cases are diagnosed in infancy or childhood, a few of them are diagnosed in adulthood.

The sweat test is the gold standard method in diagnosis. When the sweat test is found to be normal or borderline in some atypical CF patients, it is recommended to search for genetic mutations [2].

In this case report, a case who was followed up and treated with the diagnosis of recurrent pneumonia and asthma since childhood and diagnosed with CF in adolescence is presented.

CASE PRESENTATION

A 19-year-old female patient was admitted to our outpatient clinic with complaints of cough, sputum, and wheezing that had been going on for about 12 years. She had a history of asthma and chronic sinusitis for seven years. She was still using a combination of inhaled steroids and long-acting beta 2 agonists. She had never smoked and had no history of pet feeding. She wasn't working any job. While breathing room air, oxygen saturation of 98%, pulse rate of 86/min, respiratory rate of 17/min, and body mass index of 22.3 kg/m² were measured by pulse oximetry. In the respiratory examination, inspiratory crackles and squawks were heard in the bilateral upper areas.

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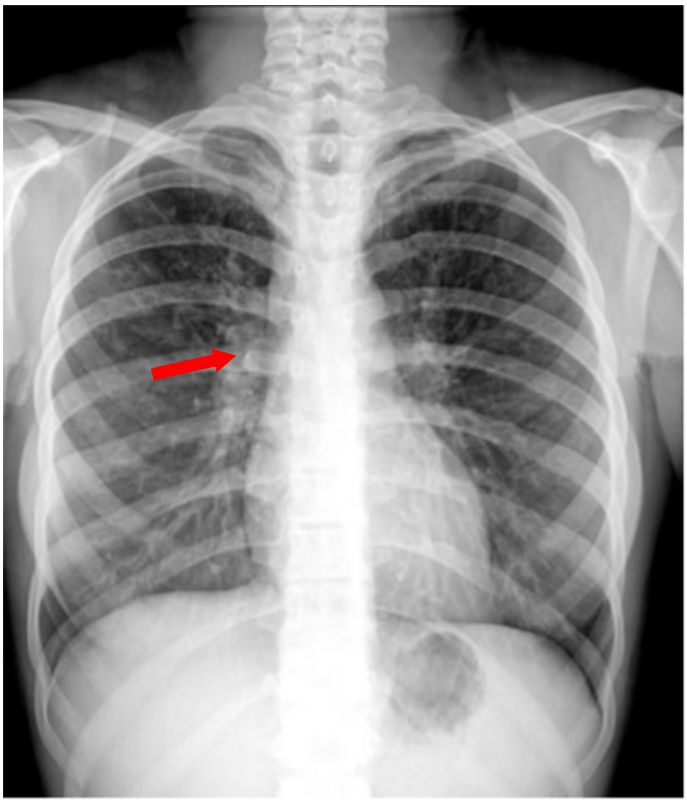

Fig. 1. Enlargement of bilateral upper-middle zones and train rail appearance on PA chest X-ray (red arrow).

Chest X-ray showed increased streaking and train rail appearance in bilateral upper-middle zones (Fig. 1). In the thorax computed tomography taken in an external center six months ago, there was centrally located bronchiectasis in the upper lobes of both lungs, accompanying peribronchial thickening and reticulonodular infiltrates (Fig. 2).

Complete blood count and biochemical parameters were normal, total IgE was 830 IU/mL. There was no growth in nonspecific sputum culture. ARB staining was negative and mycobacterial culture was negative. In the pulmonary function test, FEV1/FVC was 71.75%, FVC: 2.70 L (74.2%), FEV1: 1.94 L (61%), and DLCO 72.6%. Skin prick test was positive for Aspergillus.

Since childhood, the patient has regularly received many inhaler steroid and bronchodilator combinations in various device forms, oral antihistamine and leukotriene receptor antagonists, antitussive drugs, empirical antibiotic treatments, intranasal steroids, oral steroid treatments, but never completely regressed in his complaints. The patient with radiologically upper lobe dominant central bronchiectasis also had a history of not gaining weight and frequent diarrhea when infancy was questioned. In line with this information, genetic testing was requested from the patient with the suspicion of cystic fibrosis. As a result of the genetic test, heterozygous mutations were detected in two genes (p.Phe508del and p.Met1354Lys) for CF. ABPA was not considered in the patient who did not meet the necessary conditions according to ISHAM criteria for allergic bronchopulmonary aspergillus (ABPA). The patient was referred with the diagnosis of cystic fibrosis to be followed up and consulted in an experienced center.

DISCUSSION

Cystic fibrosis is an autosomal recessive inherited disease caused by pathological variants in the CFTR gene located on the long arm of chromosome 7. It is common in the white race. Its frequency is one in 2000 to 3000 live births [3]. Although its frequency is not known clearly in our country, it is considered a rare disease. However, considering consanguineous marriages in our country increase the incidence of this disease with OR transition [4].

The sweat test is the gold standard method in diagnosis. A sweat chloride concentration of > 60 mmol/L is diagnostic for CF. The sweat test may be normal or borderline in some atypical CF patients. It

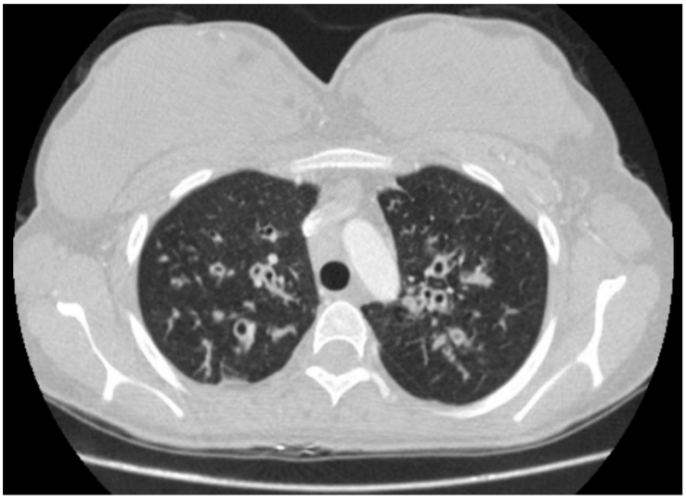

Fig. 2. Centrally located bronchiectasis in the upper lobes of both lungs; accompanying peribronchial thickening and reticulonodular infiltrates on thorax computed tomography.

is recommended to investigate genetic mutations in these patients [2]. The most common type of mutation in the CFTR gene is the deltaF508 mutation, which is 66% common worldwide. However, it has been determined that there are about 2000 mutations of the CFTR gene apart from deltaF508. Different mutations in the CFTR gene cause the disease to occur with different pictures [4].

The early diagnosis affects the life expectancy and quality of patients. Newborn screening was included in the screening program in our country on January 1, 2015. The measurement of immunoreactive trypsinogen (IRT) in a postpartum heel blood sample is used for screening purposes. In case of high values, sweat tests and gene mutation analysis are performed in the follow-up [5].

The disease mainly affects the upper and lower respiratory tract, pancreas, gastrointestinal tract, reproductive organs, and exocrine sweat glands. Chlorine and bicarbonate transport in the epithelial tissue is disrupted, leading to a decrease in moisture in lumen secretions and intensification of secretions [3].

Meconium ileus, prolonged neonatal jaundice, growth retardation, chronic cough, recurrent pneumonia, and recurrent bronchiolitis are seen in infancy. While most of the cases are diagnosed in infancy, patients with the atypical clinical course can be diagnosed in adolescence or adulthood [2]. Our patient was a 19-year-old patient who had asthma, bronchiectasis, and chronic sinusitis since childhood. According to her sister's description, the inability to gain weight in infancy and retardation in development were stated, but this story could not be fully confirmed since the patient was not raised by her parents.

Chronic endobronchial infection and progressive airway obstruction in the early respiratory system lead to impaired airway function. Late complications result in bronchiectasis, respiratory failure, and premature death. Adolescents and adults present with chronic cough and sputum, idiopathic bronchiectasis, chronic sinusitis, nasal polyposis, exercise intolerance, dyspnea, and recurrent pneumonia. Lung complications are the main cause of morbidity and mortality [6]. Our patient had a history of antibiotic use due to recurrent pulmonary infections in the early period, treatment with recurrent wheezing and asthma, and bronchiectasis detected within the last year.

The main goals of treatment are to protect respiratory functions and quality of life and to increase life expectancy. Because lung disease is the leading cause of morbidity and mortality, maintaining respiratory health is the main focus of cystic fibrosis treatment. Airway clearance techniques, mucolytics, treatment of infections with appropriate antibiotics, treatment of the host's inflammatory response with anti-inflammatory drugs to delay airway obstruction, bronchodilator and inhaler steroids in patients with asthma, and steroid therapy in patients with ABPA are the main targets. In addition, caloric support and pancreatic enzyme replacement are required in patients with inadequate exocrine pancreatic functions [6].

In recent years, modulator therapies, which are a new treatment method, aim to improve the production and functions of the CFTR protein. To date, four modulator therapies (ivacaftor, lumacaftor/ivacaftor, tezacaftor/ivacaftor, and tezacaftor/ivacaftor/elexacaftor) have been approved for use in the treatment of patients with specific CFTR mutations [7].

CONCLUSION

Although most of the patients are diagnosed in infancy or childhood, it may have a clinical atypical course in cases where the CFTR protein partially functions. Lung findings may occur at a later stage or CF is not considered due to this atypical course. Our patient was also diagnosed with CF at an adolescent age. CF should be considered in the etiology of all adolescents and adults with bronchiectasis and unexplained signs of chronic lung disease.

Authors' Contribution

Study Conception: MY; Study Design: MY, LCM; Supervision: MY, LCM; Funding: N/A; Materials: MY; Data Collection and/or Processing: MY, LCM; Statistical Analysis and/or Data Interpretation: MY; Literature Review: MY; Manuscript Preparation: MY and Critical Review: MY.

Informed Consent

Written informed consent was obtained from the patient for publication of this case and any accompanying images or data.

Conflict of interest

The authors disclosed no conflict of interest during the preparation or publication of this manuscript.

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