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Young's syndrome, a rare syndrome that can cause infertility and mimics cystic fibrosis and immotile-cilia syndrome: a case report

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Abstract. – INTRODUCTION: Young's syndrome (YS) is a rare, inherited syndrome commonly seen in middle-aged men with chronic rhinosinusitis, nasal polyps, decreased fertility due to azoospermia, and bronchiectasis. In this paper, we present a case of YS of unknown cause together with a literature review.

CASE PRESENTATION: A 28-year-old male patient with the complaints of cough, sputum, recurrent nasal congestion, and shortness of breath lasting for more than ten years, was admitted to our clinic after bronchiectasis was observed in the thoracic computed tomography.

CONCLUSIONS: An accurate diagnosis of YS is usually made late, which reduces patients' quality of life and leads to chronic respiratory problems. Failure to diagnose this disease may expose the patient to unnecessary and repeated hospitalizations and examinations, and result in treatment failure.

Key Words:
Young's syndrome, Bronchiectasis, Nasal polyp, Azoospermia.

Introduction

When diagnosing Young's syndrome (YS), also called sinusitis-infertility syndrome, a complete history should be taken first and a physical examination should be then performed. The diagnosis of YS is based on the presence of chronic sinopulmonary infections, persistent azoospermia, normal spermatogenesis, and characteristic epididymal findings, as well as the exclusion of cystic fibrosis (CF) and immotile-cilia syndrome (ISS) with similar symptoms to YS[1,2]. YS is more common in men, while CF occurs equally in both genders[3].

In addition to the unknown estimated prevalence of YS, its pathophysiology has also not been clarified. Childhood mercury exposure and genetic etiologies have been suggested. Since the introduction of restrictions on mercury use, the incidence of YS has also decreased[4].

Case Presentation

A 28-year-old male patient, who was selling vegetables and fruits in a market, presented to the emergency department of our hospital with the complaints of cough, sputum, wheezing, fever, chills, chills, headache, and chest pain. He was referred to our clinic due to widespread cystic bronchiectasis detected on the thoracic computed tomography (TCT) taken in the emergency department and a low blood partial oxygen level. We admitted the patient to our clinic for treatment and further examination. When investigating the etiology of bronchiectasis, his anamnesis revealed that the patient had a consanguineous marriage, had a febrile lung disease once in his childhood, frequently presented to the ear, nose and throat outpatient clinic due to sinusitis complaints, and underwent an operation with the diagnosis of bilateral nasal polyps six months earlier. There was no similar diseases in his family.

In his physical examination, the patient was observed to be cachectic, conscious, oriented, and cooperative but had poor self-care. His blood pressure was 110/70 mmHg, pulse rate was 90 beats per minute, respiratory rate was 22 breaths per minute, and body temperature was 37.5°C. His arterial oxygen saturation (SaO_2) was 91% on room air. In the respiratory system examination, coarse crackles and bilateral diffused rhonchi were heard in both basal lung areas. The patient's complete blood count tests were normal. Serum

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immunoglobulin level was Ig A: 2.45 g/L, Ig M: 1.03 g/L, Ig G: 18.19 g/L, Total Ig E: 145 IU/ml. Alpha-1 antitrypsin level 161 mg/dl and cystic fibrosis genetic tests were normal. Fungi and acid-fast bacillus were also negative in four sputum samples. Oral flora cells grew in the sputum culture. On TCT, significant changes consistent with diffused cystic-cylindrical bronchiectasis were observed in both lungs (Figure 1). Pulmonary function tests (PFT) were forced expiratory volume in 1 (FEV_1) 21%, force vital capacity (FVC) 26%, FEV_1/FVC 67%. Obstruction was detected in PFT, and the result of the reversibility test was positive. Inhaled bronchodilator therapy was added to the patient's treatment. YS was considered, but it was known that the patient was the father of two children. Urology clinic consultation was requested for the investigation of infertility due to azoospermia, which is known to be associated with YS. Spermogram, scrotal doppler ultrasonography and sex hormone tests were requested, and the patient was diagnosed with azoospermia by the urology specialists based on the results. The diagnosis of YS was finalized based on sinopulmonary infection, bronchiectasis, and azoospermia.

The patient was treated with 400 mg of moxifloxacin antibiotic, acetylcysteine mucolytic, chest physiotherapy and inhaled budesonide, ipratropium bromide-salbutamol in our clinic for seven days. Influenza and pneumococcal vaccines were recommended to the patient whose vaccines were missing. After the treatment, clinical improvement was observed, and necessary information was given to the patient. Follow-up visits were scheduled, and the patient was discharged.

Discussion

In order to professionally undertake the evaluation, management and treatment of patients with YS, a correct diagnosis must first be made. When making the diagnosis, the first step should be to exclude Kartagener's syndrome (KS), the most common presentation of ISS, and CF with a known genetic transmission.

CF is an autosomal recessive inherited disease characterized by chronic lung disease, pancreatic insufficiency, intestinal obstruction, male infertility, and an increased chloride in sweat. The disease develops due to mutations in the cystic fibrosis transmembrane regulator (CFTR) gene[5]. Patients with YS usually lead an active life with a normal life expectancy. The rate of lung function decline is slower in YS than in CF[6].

According to our review of the literature, a total of 55 cases of YS have been previously reported[1,7-11]. The mean age of 19 patients was 34.4 years, three patients were aged between 30 and 48 years, and 29 patients were 31.3 ± 0.8 years. Three patients are not available. According to the data obtained, chronic bronchitis was observed in two patients, bronchiectasis in 23 patients, and acute/chronic sinusitis in 22 patients. All the 55

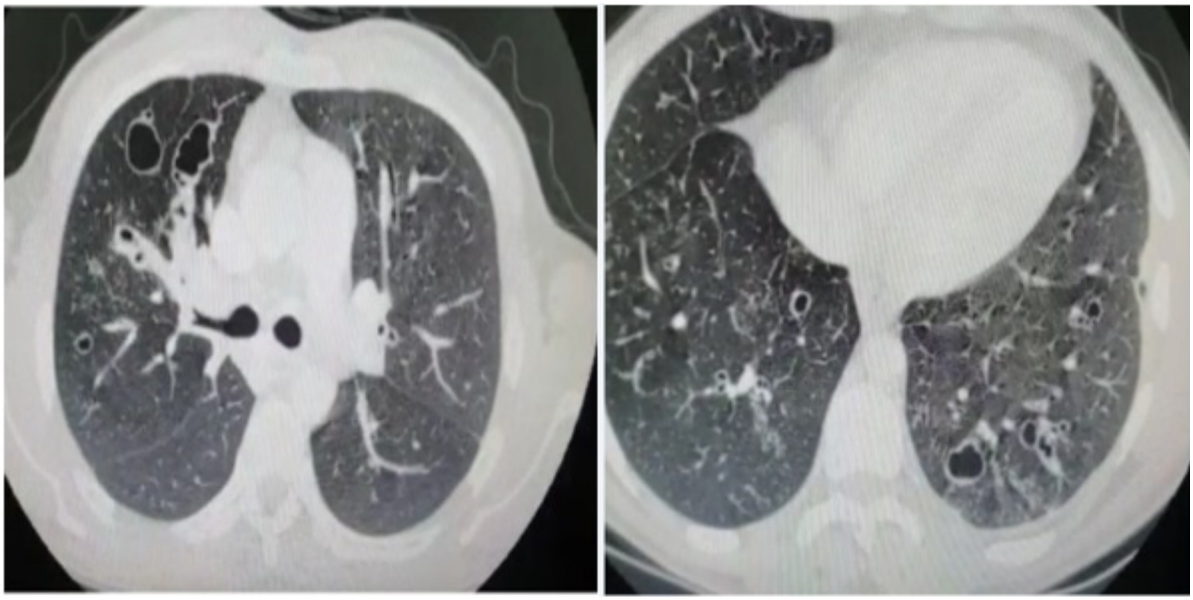

Figure 1. Bilateral cystic bronchiectasis detected on the thoracic computed tomography of the patient.

patients had obstructive azoospermia. In one of these reviewed studies[1] including 29 patients, infertility was observed in 22 patients and fertility subsequently developed in the remaining five. In another case report[8], a 28-year-old patient was diagnosed with YS while being investigated for infertility, and his wife became pregnant after a successful epididymostomy operation. In the literature, dextrocardia, which is similar to KS, has been described in two patients with YS[8,9]. While mercury is implicated in the etiology of YS, there has also been a study[9] describing three cases of YS in a country where calomel has been banned since 1960. In another study[10] comparing four patients with YS and five patients with PSD, the authors stated that transmission electron microscopy was a useful technique for evaluating ciliary abnormalities in nasal mucosa samples. In a study[11] of 13 patients, it was determined that mild respiratory tract infections were present in patients with YS, none had complicated infections, such as *P. aeruginosa* and *Staphylococcus aureus*, and there were mild decreases in PFT. In the same study, the FEV_1 second was normal or there was only a slight decrease in most patients. In patients with YS, the incidence of CFTR mutations did not significantly differ from the expected carrier frequency.

Conclusions

The possibility of YS should always be kept in mind in patients presenting with recurrent upper and lower respiratory tract infections, sinusitis, or bronchiectasis. Although there is no easy, reliable and non-invasive diagnostic test or specific treatment for this disease, failure to recognize it may expose the patient to unnecessary and repeated hospitalizations, examinations, and inappropriate treatments.

Conflicts of Interest

The Authors declare that they have no conflict of interests.

Informed Consent

Written consent was obtained from the patient.

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